and others), it was reported that 29% of the infants exhibited normal movement patterns, and two of these infants were SMA patients who had received treatment. In a study examining a case of Dejerine–Sottas syndrome, it was reported that both prenatal ultrasound and postnatal GMA revealed decreased movement variety, as well as abnormal quality of the GMs and isolated leg movements. In conclusion, although the current literature is limited, the studies reviewed suggest that many infants with NMD exhibit spontaneous movement abnormalities. However, disease-specific movement patterns are not yet clear. Further studies are needed to better understand early spontaneous motor behavior in NMD.

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SMA – PHYSIOPATHOLOGY 1.98eP-199eP; 1.83P-1.87P

1.98eP

Investigation of long non-coding RNA profiles derived from peripheral blood mononuclear cells in spinal muscular atrophy

Ayse Ipek Polat [1], Uluc Yis [1], Semra Hiz [1], Yavuz Oktay [3], Aslı Süner Karakülah [2], Gamze Sarıkaya Uzan [1], Cagatau Gunay [1], Didem Soydemir [1], Özlem Özsoy [1]
[1] *Dokuz Eylül Üniversitesi, Faculty of Medicine, Division of Pediatric Neurology, Izmir, Turkey;* [2] *Ege University, Faculty of Medicine, Dept Biostatistics and Medical Informatics, Izmir, Turkey;* [3] *Izmir International Biomedicine and Genome Center, Izmir, Turkey*

Spinal muscular atrophy (SMA) has been detectable through newborn screening programs since May 2022, enabling patients to access early treatment. However, during the newborn period, cases are often asymptomatic, making it difficult to predict which SMA subtype will develop. Even among patients who receive the same treatment opportunities, variable therapeutic responses (complete, partial, or no response) are observed. With this proposed project, we aim to minimize morbidity by enabling early identification of SMA subtypes, overcome the challenges in defining clinical subtypes following the advent of new therapies, and investigate the potential role of long non-coding RNAs (lncRNAs) as biomarkers. The main research question of our study is: "Does Spinal Muscular Atrophy lead to alterations in lncRNA levels, and can these changes be utilized to develop a diagnostic tool for determining SMA subtypes in asymptomatic individuals identified through screening?" In our study, long non-coding RNA (lncRNA) expression levels obtained from peripheral blood mononuclear cells (PBMCs) of SMA patients and healthy controls will be analyzed and compared. SMA is an anterior horn cell disease that leads to a neurodegenerative clinical course if left untreated. We aim to identify changes in lncRNA expression across SMA and its subtypes, as well as to evaluate the effects of treatment on these changes. Furthermore, PBMC-derived lncRNA profiles will be analyzed across different SMA clinical subtypes to provide insights into early subtype determination, prognosis, and disease progression. This approach aims to facilitate early identification of SMA subtypes before the onset of clinical manifestations and to improve the monitoring and follow-up of patients whose clinical course may change with various treatment modalities. Peripheral blood samples will be obtained from patients presenting with clinical signs and symptoms of SMA at the Dokuz Eylül University Dept Pediatric Neurology, as well as from healthy children without chronic diseases during routine outpatient follow-ups. From these samples, only 2 mL of blood will be allocated for this study, and isolation procedures will be performed on the same day. Peripheral blood mononuclear cells (PBMCs) will be isolated using density gradient centrifugation, followed by RNA extraction from these cells. Using the obtained data, differences in lncRNA expression between healthy controls and SMA patients will be evaluated.

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1.99eP

Neurofilament study: assessment of neurofilament levels in premature newborns

Marie Machiels [1], Tamara Dangouloff [2], Nadège Hennuy [2], Vincent Rigo [2], Laurent Servais [2,3]
[1] *University of Liège, Belgium;* [2] *University Hospital and University of Liège, Belgium;* [3] *University of Oxford, UK*

Recent therapeutic advances in spinal muscular atrophy significantly changed the natural history of the disease. Early administration of disease modifying treatments has demonstrated better efficacy when delivered as early as possible, ideally before the first symptoms. In this context, biomarkers such as neurofilaments offer the opportunity to document neuronal death before the first symptoms appear and are therefore important to guide pre-symptomatic intervention. We recently used neurofilament as a surrogate biomarker to demonstrate the interest of elective pre-term caesarean section in a baby with 2 copies of SMN2 identified in utero. Unfortunately, no reference values currently exist for neurofilaments in preterm infants without neurological disease, although some SMA forms are symptomatic in the neonatal period. Our project aims to establish normative neurofilament values in preterm newborns between 34 and 37 weeks and to study their postnatal evolution. We are conducting a prospective observational study including 15 participants (5 born at 34 weeks, 5 at 35 weeks, and 5 at 36 weeks). A 1 ml umbilical cord blood sample is collected at birth, with additional samples taken during scheduled blood tests at 35, 36, or 37 weeks. Analyses focus on comparing neurofilament concentrations by gestational age, their postnatal trajectory, and comparison with published values in SMA patients. The study began on March 13, 2026, and is planned for one year. We have currently enrolled one patient born at 36 weeks, for whom three samples have been collected. We will present the baseline data at the WMS.

http://dx.doi.org/10.1016/j.nmd.2026.106680

1.83P

From CSF proteomics to functional models: validation of SMA biomarkers in C. elegans

Pamela Santonicola [1], Chiara Panicucci [2], Giada Onorato [1], Maria Bolla [1], Chiara Fiorillo [3,6], Delia Gagliardi [4], Stefania Corti [4], Angela Chambery [5], Elia Di Schiavi [1], Claudio Bruno [2,6]
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Biomarker validation is a critical step in translating molecular findings into clinically relevant tools for disease stratification and monitoring. We previously identified CNTN1 and NRXN3 as potential biomarkers to distinguish severe SMA type 1 from milder forms through cerebrospinal fluid (CSF) proteomic profiling of treated and untreated SMA patients. Also, serum CNTN1 levels were measured in SMA type 1–3 patients and were significantly reduced only in SMA type 1 compared to controls. Here, we investigated the expression and modulation of the orthologs of CNTN1 and NRXN3 in C. elegans models of SMA. The C. elegans orthologs of CNTN1, rig-6, and of NRXN3, nrx-1, were analyzed in the severe SMA model (smn-1 knockout, allele ok355) using RT-qPCR. We observed a significant downregulation of rig-6 ($p<0.01$) in the KO, alongside an upregulation of nrx-1 ($p<0.05$), consistent with patient-derived data. Given the known role of SMN1 in RNA splicing, we further assessed alternative splicing of exon 24 in nrx-1 isoforms (ex24 and Δex24) via semi-quantitative RT-PCR. A significant increase in the exon-included isoform was detected in the KO ($p<0.05$), indicating nrx-1 splicing alterations associated with SMN deficiency. To explore the functional relevance of nrx-1 modulation, we employed C. elegans genetics to generate a pan-neuronal overexpression of nrx-1. This was combined with motoneuron-specific smn-1 silencing to evaluate potential exacerbation of disease phenotypes. Neurodegeneration was assessed by fluorescent neuronal markers in vivo, while locomotion was quantified by body bend assays. The overexpression of nrx-1 did not significantly alter neurodegeneration or locomotor defects in the SMA model, suggesting that its dysregulation may represent a biomarker rather than a direct driver of pathology. Overall, our findings validate the conservation of SMA-associated molecular alterations in C. elegans and support the use of this model for biomarker validation studies.

http://dx.doi.org/10.1016/j.nmd.2026.106681

1.84P

Cell-free plasma proteomics reveal multi-dimensional biomarker signatures in spinal muscular atrophy: insights from the RHU SMART cohort

Thomas Chetot [2], Arnaud Jacquier [1,2], Mazelin Laetitia [2], Zelia Chauvaud [2], Laurent Servais [3], Shams Ribault [1,2], Laurent Schaeffer [1,2], Carole Vuillerot [1,2]
[1] *Hospices Civils De Lyon, Lyon, France;* [2] *PGNM, Université Claude Bernard Lyon 1, Lyon, France;* [3] *Oxford University, Oxford, UK*

Despite transformative disease-modifying therapies, robust biomarkers for disease monitoring and therapeutic response remain an unmet need in SMA. This study aimed to identify cell-free proteomic biomarkers in plasma of SMA patients across clinical subtypes, genotypes, and treatment modalities using high-throughput proteomics (Olink Explore HT, >5,400 proteins). Plasma samples from 26 SMA patients (types 1–3; SMN2 copies 2–4; Nusinersen, Onasemnogene abeparvovec, or untreated) were analysed, with up to 5 longitudinal timepoints

WMS 2026 Full Programme

Version: 10/09/26

WMS 2026 Poster Sessions, Short Oral Presentations, Clinical Trial Updates and Industry Symposia are not EACCME accredited. Details of these sessions are in the full Congress programme appendix.

Programme Key:

INV	**Invited Speaker**
O/LBO	**Selected Oral Presentation/Late Breaking Selected Oral Presentation**
SP	**Short Oral Presentation**
P/LBP	**Poster Presentation/Late Breaking Poster Presentation** (on display at the venue and on the virtual platform)
eP	**ePoster Presenter** (on display at the venue and on the virtual platform)
VP/LBVP	**Virtual Poster Presentation/ Late Breaking Virtual Poster Presentation** (on display on the virtual platform and on ePoster boards at the venue, outside of the scheduled poster sessions)
ICP	**Interesting Case Presentation**

Please note, all times stated in the programme are in local Hiroshima, Japan time (JST).

Monday 28 September 2026

08:30-19:00	**Pre-Congress Teaching Course** (separate registration required) *Orchid Ballroom 3, ANA Crowne Plaza*

Tuesday 29 September 2026 Click here for details of Industry Symposia 1 & 2

08:30-11:30	**Pre-Congress Teaching Course** (separate registration required) *Orchid Ballroom 3 and Acacia, ANA Crowne Plaza*	
10:00-15:00	**WMS Executive Board Meeting** (by invitation only) *ANA Crowne Plaza*	
13:00-17:00	**Registration and poster set up** *International Congress Center Hiroshima (ICCH)* *Poster set up is only available for posters being shown in the Conference Management Room*	
14:00-15:00	**Early to mid-career peer networking session** (separate registration required) *Cosmos ICCH*	
15:00-16:00	**Regional networking** (separate registration required) *Ran ICCH*	
16:00-17:00	**Industry Symposium 1** *Himawari Hall ICCH*	**Industry Symposium 2** *Phoenix Hall ICCH*
17:00-18:00	**Registration for Opening Ceremony and Networking Reception** *Level 3 Foyer, ANA Crowne Plaza*	
18:00-18:45	**Opening Ceremony** (separate registration required) *Orchid Ballroom, ANA Crowne Plaza*	
18:45-21:00	**Networking Reception** (separate registration required) *Orchid Ballroom, Acacia and Cattleya rooms, ANA Crowne Plaza*	

Wednesday 30 September 2026

Click here for full details of Industry Symposia 3-7, Poster sessions 1-2 and Short Oral presentations 1-6

All subsequent sessions will take place at ICCH
(except NMD Editorial Board Meeting and Industry Symposia 6 and 7 at ANA Crowne Plaza)

07:15-20:00	**Congress desk open**	
07:45-08:45	**Industry Symposium 3** *Himawari Hall*	**Industry Symposium 4** *Phoenix Hall*
08:50-09:30	**Congress Welcome -** Message from the President *Phoenix Hall*	
09:30-11:00	**Topic 1: Myotonic disorders (DM1), basic biology and treatment options** *Phoenix Hall* *Moderators: Vishnu Venugopalan Yamuna, All India Institute of Medical Sciences, India and Nicol Voermans, Radboud University Medical Centre, Netherlands* 09:30-10:00 **01INV Unstable DNA in myotonic dystrophy type 1: dynamics, consequences and therapeutic opportunities** ***Darren Monckton***[1] [1]*University of Glasgow, Glasgow, United Kingdom* 10:00-10:30 **02INV Brain and heart involvement in myotonic dystrophies: causes and consequences** ***Cornelia Kornblum***[1,2] [1]*Theodor-Wenzel-Werk e.V., Department of Neurology, Berlin, Germany,* [2]*University Hospital Bonn, Center for Neurology, Bonn, Germany* 10:30-10:45 **01O Different cognitive profiles and their brain MRI correlates in adult myotonic dystrophy type 1** ***Jean-Baptiste Davion***[1]*, Céline Tard, Romain Viard, Renaud Lopes, Loren Fragoso, Luc Defebvre, Grégory Kuchcinski, Xavier Delbeuck* [1]*Department of Neurology, CHU Lille, F-59000, Lille, France, Lille, France* 10:45-11:00 **02O Methylation of CCG variant repeats is associated with heterogeneous CpG methylation flanking DMPK expansion in patients with myotonic dystrophy type 1** ***Jovan Pesovic***[1]*, Pavel Bashtrykov, Milica Poljicak, Lana Radenkovic, Igor Davidovic, Vladimir Tomic, Nemanja Radovanovic, Milos Brkusanin, Vidosava Rakocevic Stojanovic, Stojan Peric, Dusanka Savic-Pavicevic* [1]*Center for Human Molecular Genetics, University of Belgrade - Faculty of Biology, Belgrade, Serbia*	
11:00-14:00	**Poster set up** *Himawari Hall*	
11:00-11:30	**Morning refreshments, exhibition and poster viewing**	
11:00-11:30	**Myology Café** Meet the committee *Sakura room*	
11:30-13:30	**Topic 1: Myotonic disorders and OPDM** *Phoenix Hall* *Moderators: Hacer Durmus, Istanbul Universitesi Capa Tip Fakultesi, Turkey and Takashi Kurashige, NHO Kure Medical Center and Chugoku Cancer Center, Japan* 11:30 -12:00 **03INV Advances in translational therapeutics for DM1** ***Masayuki Nakamori***[1] [1]*Yamaguchi University, Ube, Japan* 12:00-12:30 **04INV Central nucleation in myotonic dystrophy: beyond classical degeneration–regeneration** ***Vanessa Todorow***[1,2,3]*, Xavière Lornage, Shinichiro Hayashi, Fiorella Carla Grandi, Zoé Clerc, Jeanne Lainé, Michel Ney, Mégane Lemaitre, Corentin Rouxel, Hanseul Oh, Mie Kato, Nobuyuki Eura, Yoshihiko Saito, Francia Victoria A. De Los Reyes, Satoru Noguchi, Benedikt Schoser, Denis Furling, Ichizo Nishino, Peter Meinke, Frédérique Rau* [1]*UCL, London, United Kingdom,* [2]*Friedrich-Baur-Institute, Munich, Germany,* [3]*National Center of Neurology and Psychiatry, Tokyo, Japan* 12:30-12:45 **03O Long-term functional improvement with zeleciment basivarsen in the phase 1/2 ACHIEVE trial** ***Karlien Mul***[1]*, Guillaume Bassez, Jordi Diaz-Manera, James Lilleker, Marika Pane, Richard Roxburgh, Benedikt Schoser, Christopher Turner, Soma Ray, Huaihou Chen, Shauna Andersson, Douglas Kerr, Valeria Sansone* [1]*Radboud University Medical Center, Beuningen, Netherlands*	

	12:45-13:00	**04O ELIXIR, a synthetic peptide modulator of sodium channel inactivation, suppresses myotonia in a mouse model of hyperkalemic periodic paralysis** *Fenfen Wu, Ryan Mwhling, Marbella Quinonez, Manu Ben-Johny, **Steve Cannon**[1]* *[1]David Geffen School of Medicine At UCLA, Los Angeles, United States*
	13:00-13:15	**05O Tandem repeat pathogenicity signature unlocks oculopharyngeal distal myopathy discoveries** *Matt Danzi, Isaac Xu, Liedewei Van de Vondel, Stefano Facchini, Riccardo Curro, Gianina Ravenscroft, Matthew Wheeler, Andrea Cortese, **Stephan Zuchner**[1]* *[1]University Of Miami, Miami, United States*
	13:15-13:30	**06O Gene-specific repeat architecture and methylation shape clinical variability in oculopharyngodistal myopathy** ***Nobuyuki Eura**[1,2], Satoru Noguchi, Megumu Ogawa, Kyuto Sonehara, Ai Yamanaka, Takashi Kurashige, Shinichiro Hayashi, Yukinori Okada, Kazuma Sugie, Ichizo Nishino* *[1]Department of Neurology, Nara Medical University, Kashihara, Japan, [2]Department of Neuromuscular Research, National Center of Neurology and Psychiatry, Kodaira, Japan*
13:30-14:30	**Lunch, exhibition and poster viewing**	
13:45-14:30	**NMD Editorial Board Meeting** (by invitation only) *Acacia Room, ANA Crowne Plaza*	
14:30-15:30	**Poster session 1** **1.009eP-1.020eP, 1.001P- 1.010P: LGMD - Clinical care and case reports** **1.021eP- 1.025eP, 1.011P- 1.014P: LGMD - Imaging and outcome measures** **1.026eP- 1.029eP, 1.015P-1.016P: LGMD - Pathophysiology of NMDs** **1.030eP- 1.031eP, 1.017P: LGMD – Treatments** **1.032eP- 1.036eP, 1.018P- 1.026P: Multiorgan syndromes** **1.037eP- 1.054eP, 1.027P- 1.033P: Neurogenic Disorders - Clinical care and case reports** **1.055eP- 1.058eP, 1.034P, 1.036P: Neurogenic disorders - Imaging and outcome measures** **1.037P-1.038P: Neurogenic disorders – Physiopathology** **1.039P-1.043P: Neurogenic Disorders – Treatments** **1.059eP, 1.044P-1.050P: Novel molecular genetic approaches for NMD** **1.060eP- 1.088eP, 1.051P-1.073P: SMA - Clinical care and case reports** **1.089eP-1.097eP, 1.074P- 1.082P: SMA - Imaging and outcome measures** **1.098eP- 1.099eP, 1.083P- 1.087P: SMA – Physiopathology** **1.100eP- 1.104eP, 1.088P-1.090P: SMA – Treatments**	
15:30-16:00	**Short Oral Presentations 1** *Ran* **Congenital myopathies, mechanisms and therapies** **SP01-SP05** *Moderator: Yukari Endo, Juntendo University, Japan*	**Short Oral Presentations 2** *Cosmos 1* **Toxic and inflammatory myopathies** **SP06-SP09** *Moderator: Olivier Benveniste, Sorbonne Université, France*	**Short Oral Presentations 3** *Cosmos 2* **DMD treatments** **SP10-SP15** *Moderator: Aoki Yoshitsugu, National Center for Neurology and Psychiatry, Japan*
16:15-17:00	**Congress Debate** *Phoenix Hall* **Can the neuromuscular field discontinue the use of animals in research?** *Moderators: Emma Rybalka, Victoria University, Australia and Francesco Saverio Tedesco, University College London, GOSH and the Francis Crick Institute, United Kingdom* **13INV** *Hidetoshi Sakurai, Center for iPS, Japan* **14INV** *Miranda Grounds, University of Western Australia, Australia* *The WMS Congress Debate has quickly become one of the most popular elements of the entire scientific programme. This year, the debate will tackle a critical and timely question that strikes at the very heart of translational science: **Can the neuromuscular field discontinue the use of animals in research?*** *For decades, animal models have served as the cornerstone of drug discovery and disease modelling. However, persistent translational gaps, where promising preclinical breakthroughs fail in human clinical trials, coupled with major regulatory shifts such as the FDA Modernization Act 2.0 and rapid advances in human iPSC technologies, 3D bioengineered tissues, and Microphysiological Systems (MPS), have brought us to an historic crossroads.*		

	Should we completely transition away from animal models in favour of human-centric, non-animal methodologies to accelerate effective therapies for patients? Or do animal models remain an irreplaceable necessity for capturing complex, systemic physiology that can vary across the lifespan, that cannot yet be fully replicated on a chip? Is a future without animal testing a logical, scientific evolution... or an unrealistic ideal? *The debate team includes world-renowned experts in muscle biology and translational research:* ***Hidetoshi Sakurai*** *(Japan), who will argue in favour of discontinuing the use of animals in research, and* ***Miranda Grounds*** *(Australia), who will be arguing against, with expert moderation by* ***Francesco Saverio Tedesco*** *(UK) and* ***Emma Rybalka*** *(Australia).* *In an effort to demonstrate differing viewpoints, as well as to educate and entertain, Hidetoshi and Miranda will intentionally present extreme positions that may not convey their actual and nuanced perspectives on this important issue.* *To round off this interactive discussion, the debaters will offer their genuine reflections on the future of disease modelling and present their insights regarding the real-world complexities of translational research.* *We encourage members of the audience to participate in the live polling and discussion. Please join us for what we are sure will be another Congress highlight!*
17:15-18:15	**Poster session 2** **2.001eP-2.002eP, 2.001P-2.003P: Becker muscular dystrophy** **2.003eP-2.009eP, 2.004P-2.012P: CMD - Clinical care and case reports** **2.013P-2.016P: CMD – Physiopathology** **2.010eP-2.028eP, 2.85eP, 2.017P-2.039P, 2.001VP: DMD - clinical care and case reports** **2.029eP-2.031eP, 2.040P-2.052P, 2.002VP: DMD - imaging and outcome measures** **2.033eP-2.045eP, 2.047eP-2.065eP, 2.067eP, 2.053P-2.088P, 2.129P: DMD – treatments** **2.068eP-2.072eP, 2.089P-2.105P, 2.003VP: Dystrophinopathies** **2.073eP-2.084eP, 2.106P-2.128P: Pathophysiology of NMDs** **2.086eP-2.090eP: Environmental Sustainability**
18:15-18:45	**Short Oral Presentations 4** *Ran* **Congenital myopathies** **SP16-SP20** *Moderator: Yuko Shimizu-Motohashi, National Center of Neurology and Psychiatry, Japan*
	Short Oral Presentations 5 *Cosmos 1* **Myotonic dystrophy mechanisms** **SP21-SP24** *Moderator: Guillaume Bassez, Pitié Salpetrière Hospital, France*
	Short Oral Presentations 6 *Cosmos 2* **Modeling disease mechanisms** **SP25-SP27** *Moderator: Helge Amthor, Versailles University, France*
19:00-20:00	**Industry Symposium 5** *Phoenix Hall (ICCH)*
	Industry Symposium 6 *Orchid Higashi, ANA Crowne Plaza*
	Industry Symposium 7 *Orchid Nishi, ANA Crowne Plaza*

Thursday 1 October 2026

All sessions will take place at ICCH

07:00-16:00	**Congress desk open**
08:00-09:00	**Early to mid-career peer breakfast session** *Myology Café, Sakura Room*
09:00-10:30	**Topic 2: Drug-induced myopathies** *Phoenix Hall* *Moderators: Yves Allenbach, Sorbonne Université, France and Wen-Chen Liang, Kaohsiung Medical University Hospital, Taiwan* 09:00-09:30 **05INV Immune-mediated necrotizing myopathy vs. Drug-induced myopathy: the central role of statins and HMGCR-autoantibodies** ***Lisa Christopher-Stine***[1] [1]*Johns Hopkins University School of Medicine, Baltimore, United States* 09:30-10:00 **06INV MADD related lipid storage myopathy: from genetic to toxic** ***Chuanzhu Yan***[1], *Bing Wen, Jingwen Xu* [1]*Shandong University, Jinan, China*

	10:00-10:15	**07O An Interdisciplinary Evaluation of the Impact of GLP-1 Receptor Agonists in individuals with Duchenne Muscular Dystrophy** *Lily Deng, Eleanor Greiner, Cuixia Tian, Nat Nasomyont, Chet Villa, Hemant Sawnani,* ***Meilan Rutter****[1,2]* *[1]Cincinnati Children's Hospital Medical Center, Cincinnati, United States, [2]University of Cincinnati, College of Medicine, Cincinnati, United States*
	10:15-10:30	**08O Motor peripheral neuropathy in patients with nitrous oxide abuse involves homocysteine toxicity through the unfolded protein response** ***Arnaud Jacquier****[1,2], Julian Theuriet, Shams Ribault, Laurent Coudert, Carole Vuillerot, Laurent Schaeffer* *[1]CNRS UMR5261 PGNM, Lyon, France, [2]Centre de Biotechnologie Cellulaire, Hospices Civils de Lyon, Bron, France*
10:30-11:00	**Morning refreshments, exhibition and poster viewing**	
10:30-11:00	**Myology Café** Meet the committee *Sakura room*	
11:00-13:00	**Topic 2: Drug-induced and infectious myopathies** *Phoenix Hall* *Moderators: Marianela Schiava, The John Walton Muscular Dystrophy Research Centre, United Kingdom and Werner Stenzel, Charité University Hospital Berlin, Germany*	
	11:00-11:30	**07INV Infectious myositis: from pyomyositis to parasitic infection** ***Jantima Tanboon****[1]* *[1]Siriraj Hospital Mahidol University, Bangkok, Thailand*
	11:30-12:00	**08INV Immune checkpoint inhibitors induced myotoxicities - from pathophysiology to personalized management** ***Joe-Elie Salem****[1]* *[1]Sorbonne University - APHP, Paris, France*
	12:00-12:15	**09O Drug-induced myopathies: a pharmacovigilance-based study of the spectrum of drug toxicity, with a focus on anticancer agents** *Mohamed-Yacine Khitri, Paul Gougis, Kevin Bihan, Bénédicte Lebrun-Vignes,* ***Julian Sanchez-Dal Cin****[1], Yves Allenbach* *[1]Assistance Publique Hôpitaux De Paris - Aphp, Paris, France*
	12:15-12:30	**10O Defining the Histopathologic Spectrum of ICI-Related Myositis: Toward Validation of a Standardized Diagnostic Score** ***Beatrice Labella****[1,2], Sami Tayb-boulahfa, Teresinha Evangelista, Hortense Chassepot, Yves Allenbach, Sarah Leonard-louis* *[1]Neuropathology Unit, APHP, Pitié-Salpêtrière Hospital, Paris, France, [2]Sorbonne Université, Paris, France*
	12:30-12:45	**11O A comparative study of sporadic and immune checkpoint inhibitor-related forms of myasthenia gravis-myositis (MG-IM) overlap syndrome** ***Antonio Lauletta****[1], Elena Rossini, Francesca Beretta, Valentina Damato, Simone Rossi, Raffaele Iorio, Enrico Marchioni, Antonio Petrucci, Alberto Vogrig, Carmela Romano, Laura Tufano, Francesca Forcina, Luca Leonardi, Stefania Morino, Silvia Falso, Gregorio Spagni, Sofia Marini, Paola Bini, Laura Fionda, Matteo Garibaldi* *[1]Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome, Italy, Rome, Italy*
	12:45-13:00	**Q&A Session**
13:00-14:00	**Lunch, exhibition and poster viewing**	
14:00-16:00	**Interesting case discussions** *Phoenix Hall* *Moderators: Jorge Bevilacqua, Hospital Clínico Universidad de Chile, Chile, Emna Farhat, The Muscle Lab, Tunisia and Wenhua Zhu, Huashan Hospital, Fudan University, China*	
16:00-18:30	**Poster viewing/group activity** (separate registration required) *Please note there is no group activity networking reception this year.*	

Friday 2 October 2026 Click here for full details of Poster Sessions 3 & 4 and Short Oral Presentations 7-12

All sessions will take place at ICCH

08:00-18:00	**Congress desk open**
08:30-10:15	**Topic 3: Trust, transparency and therapeutic understanding** *Phoenix Hall* *Moderators: Tamara Dangouloff, University of Liege, Belgium and Teerin Liewluck, Mayo Clinic Rochester, United States of America* 08:30-08:45 **12O Raising reasonable expectation : A collective responsibility** *Laurent Servais, President, WMS* 08:45-09:15 **09INV Hope You Can Trust: Transparency as the Foundation of Breakthroughs in Treating Neuromuscular Disorders** *Emily Reuben, Duchenne UK, United Kingdom* 09:15-09:45 **10INV From trials to treatment: Risk–benefit misestimation across the therapeutic lifecycle** *Kathryn Wagner, Johns Hopkins School of Medicine, United States* 09:45-10:15 **11INV Data, disclosure, and the meaning of benefit in NMD trials** *Maryam Oskoui, Centre Hospitalier Universitaire Sainte-Justine, Canada.*
10:15-10:45	**Morning refreshments, exhibition and poster viewing**
10:15-10:45	**Myology Café** Meet the committee *Sakura room*
10:45-12:30	**Topic 3: Advancements in neuromuscular diseases** *Phoenix Hall* *Moderators: Duygu Selcen, Mayo Clinic, United States of America and Conrad Weihl, Washington University Medical University, United States of America* 10:45-11:15 **12INV Developing clinical trials for mitochondrial diseases** ***Rita Horvath***[1] [1]*University of Cambridge, Cambridge, United Kingdom* 11:15-11:30 **13O Myo-guide: a one-stop cloud platform for AI-based muscle MRI segmentation, quantification, analysis and sharing** ***José Verdú Diaz***[1,2,3], *Longdan Hao, Carla Bolaño-Díaz, Sam Fitzsimmons, Natalia Kaliteevskaya, Alejandro Gonzalez Chamorro, Stephen Wandera, Sipho Ndlovu, Joel Mannion, Michele Croce, Libby Grey, Holly Borland, Kieren Hollingsworth, Michela Guglieri, Chiara Marini Bettolo, Volker Straub, Giorgio Tasca, Jaume Bacardit, Jordi Diaz-Manera* [1]*John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle upon Tyne, United Kingdom,* [2]*NIHR Newcastle Biomedical Research Centre, Biomedical Research Building, Campus for Ageing and Vitality, Newcastle upon Tyne, United Kingdom,* [3]*Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom* 11:30-11:45 **14O Design of skeletal muscle-specific promoters active in slow and fast muscle fiber types for rAAV-driven treatment of LGMDR1** ***Ruby Goldstein De Salazar***[1], *Irina Kramerova, Diana Becerra1, Bradley Smith, Stephen Hauschka, Jeffrey Chamberlain, Melissa Spencer* [1]*UCLA, Los Angeles, United States* 11:45-12:00 **15O VMA21 deficiency leads to autophagic dysregulation, altered vesicle trafficking, and complement deposition in X-linked myopathy with excessive autophagy** ***Michio Inoue***[1], *Christian A. Suarez, Sara K. Pittman, Eileen M. Lynch, Andrew Moran, Teresinha Evangelista, Conrad C. Weihl* [1]*Washington University in St. Louis, St Louis, United* 12:00-12:15 **16O Bi-allelic variants in DMXL1 cause a novel autophagic vacuolar myopathy** ***Chiara Folland***[1], *Ana Töpf, Christina Vo-Monahan, Jonathan Rodgers, David Stroud, Ichizo Nishino, Thomas Robertson, Gianina Ravenscroft* [1]*Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia, Perth, Australia* 12:15-12:30 **Q&A session**
12:30-14:00	**Lunch, exhibition and poster viewing**
12:45-13:45	**WMS General Assembly** *Phoenix Hall* **/ poster viewing for non-members**

14:00-14:30	**Sponsor meeting** *Ran* (by invitation only)		
14:15-15:15	**Poster session 3** **3.001eP-3.005eP, 3.001P-3.012P: Distal myopathies and OPMD** **3.006eP-3.015eP, 3.013P-3.020P: Drug-induced and infectious myopathies** **3.016eP-3.024eP, 3.021P-3.028P: FSHD - Clinical care and case reports** **3.025eP-3.029eP, 3.029P-3.038P: FSHD - Imaging and outcome measures** **3.030eP, 3.039P-3.047P: FSHD - Pathophysiology of NMDs** **3.031eP, 3.048P-3.049P: FSHD – Treatments** **3.032eP-3.036eP, 3.050P-3.057P: Glycogenoses** **3.037eP-3.066eP, 3.058P-3.098P: Idiopathic inflammatory myopathies** **3.067eP-3.068eP: Lipid storage myopathies** **3.069eP-3.075eP, 3.099P-3.106P: Mitochondrial disorders** **3.076eP-3.078eP, 3.107P-3.116P: Vacuolar myopathies**		
15:15-15:45	**Short Oral Presentations 7** *Ran* **Congenital muscle diseases** **SP28-SP31** *Moderator: Keiko Ishigaki, Tokyo Women's Medical University, Japan*	**Short Oral Presentations 8** *Cosmos 1* **Myotonic dystrophy therapies** **SP32-SP35** *Moderator: Benedikt Schoser, Friedrich-baur-institut Lmu München, Germany*	**Short Oral Presentations 9** *Cosmos 2* **Clinical care** **SP36-SP39** *Moderator: Kristin Ørstavik, Oslo University Hospital, Norway*
15:45-16:45	**Poster session 4** **4.001eP-4.010eP, 4.001P-4.007P: CMyo - Clinical care and case reports** **4.011eP-4.014eP, 4.008P-4.020P: CMyo - Clinical physiopathology** **4.015eP-4.017eP, 4.021P-4.022P: Congenital myasthenic syndrome** **4.018eP-4.027eP, 4.023P-4.029P: Muscle imaging** **4.028eP-4.037eP, 4.030P-4.033P: Myasthenia gravis** **4.039eP-4.059eP, 4.034P-4.070P: Myotonic disorders and Periodic Paralysis** **4.060eP-4.088eP-4.071P-4.092P: Registries, networks and care**		
16:45-17:15	**Short Oral Presentations 10** *Ran* **Neurogenic disorders** **SP40-SP43** *Moderator: Heike Kölbel, Universitätsmedizin Essen, Germany*	**Short Oral Presentations 11** *Cosmos 1* **LGMD** **SP44-SP48** *Moderator: Motoi Kanagawa, Ehime University, Japan*	**Short Oral Presentations 12** **Novel approaches in NMD** *Cosmos 2* **SP49-SP53** *Moderator: Jaakko Sarparanta, Folkhälsan Research Center, Finland*
18:30-23:00	**Networking Dinner** *Grand Prince Hotel Hiroshima* (separate registration required)		

Saturday 3 October 2026 Click here for full details of Clinical Trial Updates

All sessions will take place at ICCH

07:15-14:30	**Congress desk open**
07:30-09:00	**Clinical trial updates** *Phoenix Hall* *Moderators: Liesbeth De Waele, University Hospitals Leuven, Belgium and Hirofumi Komaki, National Center of Neurology and Psychiatry, Japan* 07:30-07:45 **17O** 07:45-08:00 **18O** 08:00-08:15 **19O** 08:15-08:30 **20O** 08:30-08:45 **21O** 08:45-09:00 **22O**
09:00-09:15	**Comfort break**
09:15-09:45	**Victor Dubowitz Lecture** *Phoenix Hall* *Moderators: Jenni Laitila, Folkhälsan Research Center, Finland and Laurent Servais, University of Oxford, United Kingdom* **15INV** My journey around the nuclear envelope *Gisèle Bonne, Institut de Myologie, France*
09:45-11:15	**Poster highlights** *Phoenix Hall* *Moderators: Marco Savarese, Samfundet Folkhälsan, Finland and Kazuma Sugie, Nara Medical University, Japan* 09:45-10:00 **23O** 10:00-10:15 **24O** 10:15-10:30 **25O** 10:30-10:45 **26O** 10:45-11:00 **27O** 11:00-11:15 **28O**
11:15-11:45	**Morning refreshments, exhibition and poster viewing**
11:45-13:15	**Late breaking news** *Phoenix Hall* *Moderators: Alan Beggs, Boston Children's Hospital/Harvard Medical School, United States of America and Anna Cho, Seoul National University Bundang Hospital, South Korea* 11:45-12:00 **01LBO** 12:00-12:15 **02LBO** 12:15-12:30 **03LBO** 12:30-12:45 **04LBO** 12:45-13:00 **05LBO** 13:00-13:15 **06LBO**
13:15-13:45	**Prize Giving Ceremony** *Phoenix Hall* Introduction to the WMS 2027 Congress, Helsinki, Finland, handover of the WMS flag and close of the Congress
13:45-14:30	**Homeward lunch**

WMS 2026 Congress programme appendix

Version: 10/09/26

Please note that WMS 2026 Poster Sessions, Short Oral Presentations, Clinical Trial Updates and Industry Symposia are **not** EACCME accredited.

Affiliations are shown only for the presenting author. The full list of affiliations for authors can be found in the Congress abstract book.

To leave the appendix and return to the main programme, please click the in the top corner of each page.

Tuesday 29 September 2026

To leave the appendix and return to the main programme, please click the in the top corner of each page.

16:00–17:00 **Industry Symposium 1** *Himawari Hall ICCH*

Details to be confirmed

16:00–17:00 **Industry Symposium 2** *Phoenix Hall ICCH*

Details to be confirmed

Wednesday 30 September 2026

To leave the appendix and return to the main programme, please click the in the top corner of each page.

07:45-08:45 **Industry Symposium 3** *Himawari Hall ICCH*

Details to be confirmed

07:45-08:45 **Industry Symposium 4** *Phoenix Hall ICCH*

Details to be confirmed

19:00-20:00 **Industry Symposium 5**
Phoenix Hall ICCH

Details to be confirmed

19:00-20:00 **Industry Symposium 6**
Orchid Higashi, ANA Crowne Plaza

Details to be confirmed

19:00-20:00 **Industry Symposium 7**
Orchid Nishi, ANA Crowne Plaza

Details to be confirmed

Poster Session 1: 1.009eP-1.020eP, 1.001P-1.010P: LGMD - Clinical care and case reports

1.009eP Studying CAPN3 variant co-inheritance patterns to gain insight into genetic landscape of calpainopathy
*Emma Balsera, Rebecca Lebon, Pape Ibrahima Faye, Sébastien Courrier, Nathalie Da Silva, Martin Krahn, Johan T. den Dunnen, Anaïs Baudot, Marc Bartoli, **Svetlana Gorokhova**[1,2]*
[1]Aix Marseille Univ, INSERM, MMG, U1251, Marseille, France., Marseille, France, [2]Department of Medical Molecular Genetics, Timone Hospital, AP-HM, Marseille, France., Marseille, France

1.010eP Comparison of muscle involvement in anoctaminopathy and dysferlinopathy
***Reoto Ueda**[1,2,3], Hiroto Azuma, Ai Yamanaka, Wakako Yoshioka, Shinichiro Hayashi, Satoru Noguchi, Kazuma Sugie, Ichizo Nishino*
[1]Faculty of Medicine, Nara Medical University, Nara, Japan, [2]Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan, [3]Department of Neurology, Nara Medical University, Nara, Japan

1.011eP Calpainopathy: clinical, electrophysiological, and genetic characterization of three Indonesian patients
*Luh Ari Indrawati, **Raehan Kusumah**[1], Dinda Riyanto, Adrian Harsono, Nurul Fadli, Winnugroho Wiratman, Astri Budikayanti, Fitri Octaviana, Manfaluthy Hakim*
[1]Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia,

1.012eP Genotype–phenotype correlation in calpain 3-related myopathy: a case series of early-onset and late-onset LGMDR1
***Toshiki Ochi**[1], Ken Ikegami, Yosuke Osakada, Ryuta Morihara, Yoshio Omote, Ichizo Nishino, Hiroyuki Ishiura*
[1]Department of Neurology, Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, Okayama University, Okayama, Japan

1.013eP Novel POMT2 variants associated with limb-girdle muscular dystrophy R14: genetic, histological and functional studies
*Xiaoqing Lyu, Guiguan Yang, **Pengfei Lin**[1]*
[1]Qilu Hospital Of Shandong University, Jinan, China

1.014eP A retrospective analysis of patients with sarcoglycanopathy from a single tertiary center
***Neslihan Ozcan**[1], Serdar Pekuz, Özlem Hergüner, Gulen Gul Mert*
[1]Cukurova University Dep of Pediatric Neurology, Adana, Turkey

1.15eP Beyond limb-girdile weakness: a single center experience with calpainopathy
*Serdar Pekuz, Neslihan Özcan, Gülen Gül Mert, **Özlem Hergüner**[1]*
[1]Cukurova University Faculty of Medicine, Pediatric Neurology, Adana, Turkey

1.016eP A 48-year-old man with limb-girdle muscular dystrophy type D1 presenting with neurogenic changes and fasciculations on electromyography: a case report
***Atsuko Tsuneyama**[1], Hotake Takizawa, Yasushi Oya, Wakako Yoshioka, Ichizo Nishino, Yuji Takahashi*
[1]Department of Neurology, National Center Hospital, National Center of Neurology and Psychiatry, Tokyo, Japan., Kodaira, Japan

1.017eP Clinical and genetic analysis of LGMD R1 caused by novel non-canonical CAPN3 splicing mutations
***Meirong Liu**[1], Xiaoke Wu, Guangyu Wang, Tingjun Dai, Qi Fang, Chuanzhu Yan*
[1]The First Affiliated Hospital Of Soochow University, Suzhou, China, 2Qilu Hospital of Shandong University, Shandong, China

1.018eP HMGCR-related limb-girdle muscular dystrophy: case report
*Gökcen Öz Tuncer, Seren Aydin, Çagri Doğan, Gülbahar Kurt Bayir, **Ayşe Aksoy**[1]*
[1]Division of Pediatric Neurology, Department of Pediatrics, Faculty of Medicine, Ondokuz Mayıs University, Samsun, Turkey

1.019eP FKRP-related limb-girdle muscular dystrophy R9 caused by a novel intronic variant
***Kjell Arne Arntzen**[1], Elisabeth Kjeldsen Buvang, Christoffer Jonsrud, Gry Hoem, Linda Mathisen*
[1]Department of Neurology and Norwegian Centre for Rare Diseases - Unit for Neuromuscular Disorders, University Hospital of North Norway, Tromsø, Norway

1.020eP Homozygous 13q12.12 contiguous gene deletion causing concurrent LGMD2C and ARSACS: a blended neuromuscular phenotype
***Emilie Groulx Boivin**[1], Catalina Maftei, Katherine Bonnycastle, Maryam Oskoui*
[1]McGill University, Montreal, Canada

1.001P Four additional families confirm the distinctive clinical, radiological, and genetic features of MAMDC2-related myopathy
***Macarena Cabrera Serrano**[1,2], Emilia Servian Morilla, Victoria Navarro, Cristina Domínguez-González, Ana Gonçalves, Rosário Santos, Christine C Bruels, Melanie C O'Leary, John C Kincaid, Paloma Martin-Jimenez, Henrique M Costa, Celia Azevedo Soares, Lynn Pais, Vijay Ganesh, Elicia A Estrella, Hart G.W Lidov, Basil T Darras, Peter B Kang, Gina Ravenscroft, Carmen Paradas*
[1]Harry Perkins Institute of Medical Research, Perth, Australia, [2]Insituto de Biomedicina de Sevilla, Sevilla, Spain

1.002P Muscle-predominant CRPPA dystroglycanopathy: crystal structure-informed genotype-phenotype analysis

Bochen Zhu[1], *Kexin Jiao, Xingyu Xia, Mingshi Gao, Wenhua Zhu*
[1]*Huashan Hospital, National Center for Neurological Disorders (NCND), Huashan Rare Disease Center, Fudan University, Shanghai, China, 2Department of Pathology, Huashan Hospital, Fudan University, Shanghai, China*

1.003P Clinical, genetic, and muscle MRI phenotyping of pediatric limb-girdle muscular dystrophy: a case series of 7 patients from Turkey

Esra Sarigecili[1], *Ahu Ekren, Ayse Selcan Koc, Habibe Koc Ucar, Tamer Celik, Yilmaz Akbas, Mustafa Kömür*
[1]*Adana Şehir Hastanesi, Çocuk Nöroloji, Adana, Turkey*

1.004P Genetic features in dysferlin gene in Japan

Toshiaki Takahashi[1], *Rumiko Izumi, Naoki Suzuki, Chikako Yaginuma, Hiroyuki Takenaka, Tomoko Totsune, Masaru Yoshioka, Toru Baba, Hideki Oizumi, Hiroyasu Tanaka, Tetsuya Niihori, Takafumi Hasegawa, Yoko Aoki, Masashi Aoki*
[1]*Department of Neurology, National Hospital Organization Sendai Nishitaga Hospital, Sendai, Japan*

1.005P Clinicopathological features of anoctaminopathy: focus on inflammatory pathology and the complement pathway

Hiroto Azuma[1,2,3], *Reoto Ueda, Ai Yamanaka, Shinichiro Hayashi, Satoru Noguchi, Kazuma Sugie, Ichizo Nishino*
[1]*Faculty of Medicine, Nara Medical University, Nara, Japan,* [2]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan,* [3]*Department of Neurology, Nara Medical University, Nara, Japan*

1.006P Natural history in LGMDR1: life expectancy and comorbidities in a Gipuzkoa cohort study

Patricia Garay Albizuri[1], *María Teresa Iglesias Gaspar, Lorea Blanco, Xabier Barrutia, Juan José Poza, Edurne de Egaña, Monica Rayon, Andrea Valls, Javier Basterrechea, Ane Zufiaurre, Jenita Immanuel, Nuria Rosell, Itxaso Martí, Ana Holgado, Olatz Etxebeste, Inés López Hernando, Pilar Camaño, Amets Sáenz, Adolfo López de Munain, Roberto Fernández Torrón*
[1]*Hospital de Donostia, San Sebastian, Spain*

1.007P Pathology-based clustering reveals necroptosis-associated subtypes linked to muscle weakness in dysferlinopathy

Anzhe Zheng[1], *Mingshi Gao, Nachuan Cheng, Jianying Xi, Wenhua Zhu*
[1]*Huashan Rare Disease Centre and Department of Neurology, Huashan Hospital, Shanghai Medical College, National Centre for Neurological Disorders, Fudan University, Shanghai, China*

1.008P Clinical trial readiness for LGMDR1: 12 months results from the CALNATHIS prospective study identify clinically meaningful readouts

Gianmarco Severa[1,2], *Christophe Alimi, Tanya Stojkovic, Céline Tard, Jean Baptiste Noury, Stéphane Degove, Sarah Souvannanorath, Sophie Olivier, Isabelle Richard, Edoardo Malfatti*
[1]*Reference Center for Neuromuscular Disorders, APHP Henri Mondor University Hospital, Créteil, France,* [2]*University Paris Est Créteil, Inserm, U955, IMRB, F-94010 , Créteil, France*

1.009P Real-world insights into limb-girdle muscular dystrophy, including subtype 2I/R9: treatment patterns, health care resource utilization, and costs

Karen Bartley, Amanda Howarth, Siwei Zhu, Fei Tang, Anna Talaga, ***Sajin Mathew***[1], *Jeffrey M. Statland*
[1]*BridgeBio Pharma, Inc, Palo Alto, CA, United States*

1.010P One exon, multiple phenotypes: functional insights and clinical variability in recessive titinopathies involving the final TTN exon

Maria Francesca Di Feo[1,2,3], *Martin Rees, Ay Lin Kho, Mark Pfuhl, Mathias Gautel, Bjarne Udd, Marco Savarese, TTN exon 364 Consortium*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Helsinki, Finland,* [3]*University of Genoa, Genova, Italy*

Poster Session 1: 1.021eP- 1.025eP, 1.011P- 1.014P: LGMD - Imaging and outcome measures

1.021eP Longitudinal behaviour of NSAD and muscle fat fraction as monitoring outcome measures in LGMD R2-DYSF related

Carla Florencia Bolaño Diaz[1], *Heather Hilsden, Emma J Robinson, Meredith K James, Laura Rufibach, Consortium Jain COS, Giorgio Tasca, Jordi Diaz Manera, Volker Straub*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom*

1.022eP Clinical trial readiness in dysferlinopathy from the 10-year, multicenter Jain clinical outcome study
Heather Gordish-Dressman[1,2], *Meredith James, Laura Rufibach, Heather Hilsden, Stephen Wandera, Emma Robinson, Fiyinfolu Adetunji, . COS Consortium on behalf of the Jain Foundation, Volker Straub*
[1]*Children's National Hospital, Washington, United States,* [2]*The George Washington University, Washington, United States*

1.023eP Systematic literature review of clinical outcomes and disease burden in limb-girdle muscular dystrophy subtype 2I/R9
Karen Bartley, Obaro Evuarherhe, Gemma Carter, Anna Talaga, ***Sajin Mathew***[1]*, Jeffrey M. Statland*
[1]*BridgeBio Pharma, Palo Alto, CA, United States*

1.024eP Clinical protocol proposal for LGMD-D2: validation in a patient's series
Corrado Angelini[1]*, Alicia,A Rodriguez*
[1]*Campus Pietro d'Abano, Padua, PADOVA, Italy*

1.025eP Muscle MRI patterns in dysferlinopathy patients: a case series
Chand Nagpal[1]*, Manfaluthy Hakim, Astri Budikayanti, Nurul Fadli, Adrian Ridski Harsono, Marcel Prasetyo, Winnugroho Wiratman, Ahmad Yanuar Safri, Dinda Larastika Riyanto, Fitri Octaviana, Luh Ari Indrawati*
[1]*Department of Neurology University of Indonesia, Jakarta, Indonesia*

1.011P Structured patterns of muscle involvement in CAV3-related myopathy revealed by whole-body CT imaging
Francia Victoria De Los Reyes[1,2]*, Shinichiro Hayashi, Yoshihiko Saito, Megumu Ogawa, Yasushi Oya, Satoru Noguchi, Ichizo Nishino*
[1]*HELP Learning Center Foundation, Inc., Naga City, Philippines,* [2]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan*

1.012P One-year functional decline in LGMDR2 dysferlin-related myopathy patients in India: a prospective study using standardised outcome measures
Sneha Sneha[1]*, Ashu Bhasian, Meredith James, Nimita Rani, Ajay Garg, Ayush Agarwal, Hina Narzari, Nilima Nilima, Rohit Bhatia, Lindsay Wilson, Robert Pitceathly, Michael Hanna, Volker Straub, Venugopalan Vishnu*
[1]*All India Institute of Medical Sciences, New Delhi, India*

1.013P Whole-body muscle imaging reveals stage-specific involvement and gender-dependent functional correlations in calpainopathy and dysferlinopathy
Ai Yamanaka[1,2,3]*, Reoto Ueda, Hiroto Azuma, Shinichiro Hayashi, Satoru Noguchi, Kazuma Sugie, Ichizo Nishino*
[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan,* [2]*Department of Genome Medicine Development, Medical Genome Center, National Center of Neurology and Psychiatry, Tokyo, Japan,* [3]*Department of Neurology, Nara Medical University, Nara, Japan*

1.014P COS-PREPARED: clinical outcome study in pre- and early-symptomatic patients with recessive dysferlinopathy - what is the impact of early stage LGMDR2 on function?
Meredith James[1]*, Emma-Jayne Robinson, Emma Grover, Heather Hilsden, Sarah Emmons, Carla Bolano Diaz, Ariele Barreto Haagsma, Heather Gordish Dressman, Ian Wilson, Laura Rufibach, Lisa Alcock, Volker Straub*
[1]*The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom*

Poster Session 1: 1.026eP- 1.029eP, 1.015P-1.016P: LGMD - Pathophysiology of NMDs

1.026eP Alpha-dystroglycan glycosylation in heterozygous FKRP carriers of LGMDR9
Anne Katrine Kjemsø Jensen[1]*, Mads Godtfeldt Stemmerik, Tue Leth Nielsen, John Vissing*
[1]*Copenhagen Neuromuscular Center, (cnmc), Rigshospitalet, Copenhagen, Denmark*

1.027eP Pathological and transcriptomic comparison of dysferlinopathy and immune-mediated necrotizing myopathy
Weiyan Hou[1]*, Shaojuan Ma, Hongran Wu, Guang Ji, Xueqin Song*
[1]*Department of Neurology, Second Hospital Of Hebei Medical University, Shijiazhuang, China*

1.028eP Calcium modulation improves motor function in a mouse model of dysferlinopathy
Jinhong Shin[1]*, Kyowon Seo, Hye Young Kim, Jaeil Choi, Sumin Kim*
[1]*Pusan National University Yangsan Hospital, Yangsan, South Korea*

1.029eP Human iPS-derived cardiomyocytes to study beta-sarcoglycanpathy
Angela Maione, Elena Sommariva, ***Carles Sanchez Riera***[1]*, Beatrice Vola, Bianca Cheli, Giulio Pompilio, Massimiliano Cerletti, François Gros-Louis, Yvan Torrente*
[1]*GFB Foundation, Talamona, Italy*

1.015P Identification of potential new substrates of FKRP
Bianca Cheli[1], *Andrea Bracaglia, Luca Madaro, Marina Bouché, Carles Sanchez Riera*
[1]*Sapienza University of Rome, Rome, Italy*

1.016P Longitudinal serum proteomics reveals biomarkers of disease progression in LGMD R2 dysferlin-related
Carla Florencia Bolaño Diaz[1], *Albert Jiménez-Requena Carrasco, Esther Fernandez Simon, Consotrium Jain COS, Cristina Al-Khalili Szigyarto, Jordi Diaz Manera*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom*

Poster Session 1: 1.030eP- 1.031eP, 1.017P: LGMD – Treatments

1.030eP Longitudinal comparison of creatinine- and cystatin C–based eGFR in LGMDR2
Marianela Schiava[1,2], *Heather Gordish Dressman, Brad Williams, John W Day, Zarife Sahenk, Roberto Fernandez Torron, Matt Harms, Alan Pestronk, John Vissing, Urvi Desai, Madoka Yoshimura, Jin-Hong Shin, Tasheen Mozzafar, Tanya Stojkovic, Elena Pegoraro, Jorge A. Bevilacqua, Montse Olive, Carmen Paradas, Volker Straub*
[1]*The John Walton Muscular Dystrophy Research Centre and Clinical Research Institute, Newcastle Upon Tyne, UK,* [2]*The Newcastle upon Tyne Hospitals NHS Foundation Trust and Newcastle University, Newcastle Upon Tyne, United Kingdom*

1.031eP Correction of aberrant splicing caused by intronic CAPN3 mutations using RNA-targeted therapeutic strategies in limb-girdle muscular dystrophy type R1
Gaoyuan Li[1], *Yunuo Guo, Guangyu Wang, Haoyang Liu, Xiaoqing Lv, Chuanzhu Yan, Pengfei Lin*
[1]*Qilu Hospital Of Shandong University, Jinan, China*

1.017P Muscle complement deposition is associated with disease severity and response to complement-targeted treatment in dysferlinopathy
Fuze Zheng[1], *Shuaikun Kang, Wei Zhang, Jianwen Deng, Yun Yuan, Zhaoxia Wang, Yu Meng*
[1]*Department Of Neurology, Peking University First Hospital, Beijing 100034, China, Peking, China*

Poster Session 1: 1.032eP- 1.036eP, 1.018P- 1.026P: Multiorgan syndromes

1.032eP Divergent limb phenotypes associated with PITX1 alterations: from lower limb malformations to Liebenberg syndrome
Berta Estévez-Arias[1,2,3], *Ayse Nur Kavasoglu, Anirban Majumdar, Sarah Smithsom, Julie Evans, Amaka Offiah, Paulino Sousa, Carlos Ortez, Andres Nascimento, Laura Carrera-García, Daniel Natera-de Benito*
[1]*Applied Research in Neuromuscular Diseases, Institut de Recerca Sant Joan de Déu, Barcelona, Spain,* [2]*Neuromuscular Unit, Department of Neurology, Hospital Sant Joan de Déu, Barcelona, Spain,* [3]*Genomics Unit – NGS, Diagnostic Laboratory Service, Hospital Sant Joan de Déu, Barcelona, Spain,*

1.033eP Expanding the phenotypic spectrum of GLDN-related disorders: survival beyond the neonatal period and genotype-phenotype insights
Berta Estévez-Arias[1,2,3], *María Justel, Pascal Joset, Chiara Gatnau, Jordi Pijuan, Anna Codina, Cristina Jou, Laura Carrera-García, Jesica Expósito-Escudero, Carlos Ortez, Andres Nascimento, Isabel Filges, Daniel Natera-de Benito*
[1]*Applied Research in Neuromuscular Diseases, Institut de Recerca Sant Joan de Déu, Barcelona, Spain,* [2]*Neuromuscular Unit, Department of Neurology, Hospital Sant Joan de Déu, Barcelona, Spain,* [3]*Genomics Unit – NGS, Diagnostic Laboratory Service, Hospital Sant Joan de Déu, Barcelona, Spain*

1.034eP Multilocus pathogenic variation in a pediatric patient: a quadruple molecular diagnosis shaping a complex phenotype
Heike Kölbel[1], *David Muhmann, Adam SL Graefe, Adela Della Marina, Nele Baier-Finkh, Frank J Kaiser, Ulrike Schara-Schmidt, Kristin Köhler, Peter N Robinson, Andreas Roos*
[1]*Universitätsmedizin Essen, Essen, Germany*

1.035eP Neonatal-onset myotonia further expanding the spectrum of SCN1A-related arthrogryposis multiplex congenita with epileptic encephalopathy and CNS involvement
Pelin Özlem Şimşek Kiper, Ülkühan Öztoprak, Çağrı Temuçin, Rahşan Göçmen, Gülen Eda Utine, Dilek Yalnızoğlu, ***Göknur Haliloğlu***[1]
[1]*Hacettepe University Faculty of Medicine, Ankara, Turkey*

1.036eP A case of severe neonatal hypotonia with IPEX myositis cured after hematopoietic stem cell transplantation
Nicolas Chrestian[1], *Zoe Labelle, Benjamin Ellezam, Louis Marois*
[1]*Centre Mère Enfant Soleil, Quebec, Canada, 2CHU de ste Justine, Montreal, Canada*

1.018P Expanding the genetic spectrum of arthrogryposis: a case series

Jasmine Chew[1], *Lein Dofash, Christina Vo-Monahan, Phillipa Lamont, Anita Cairns, Margit Shah, Esther Tantsis, Zachary McPherson, Lyndal Douglas, Joshua Back, Melanie Wong, Matthias Axt, Richard Webster, Gianina Ravenscroft*
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1.019P When rarity deceives: three genetic disorders masquerading as myopathy — diagnostic pitfalls from a neuromuscular clinic

Gauthier Remiche[1], *Laurence Desmyter, Sandra Coppens, Sophie Lecomte, Karin Dahan, Rosy N'Gbo N'Gbo Ikazabo, Pierre Carlier, Afarine Madani, Isabelle Vandernoot*
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1.020P Pyridostigmine in PURA syndrome beyond infancy: a case series

Tania Dafer[1], *Emilie Groulx-Boivin, Daniel Charouf, Elisabeth Simard-Tremblay, Nicolas Chrestian, Chantal Poulin*
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1.021P ALMS1 mutation disrupts primary cilium structure and muscle stem cell function in Alström syndrome

Ana Saric[1], *Lola Albertella, Baptiste Periou, Olivier Conscience, Edoardo Malfatti, Cyril Gitiaux, François Jérôme Authier, Ludovic Martin*
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1.022P Beyond contractures: neurological and neuromuscular findings in arthrogryposis multiplex congenita

My Vuong Hermansen[1,2,3], *Kristin Ørstavik, Unni Steen, Mathias Toft, Cathrine Brunborg, Lena Lande Wekre*
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1.023P A dominant SPTAN1 missense variant associated with congenital malformation and myopathy syndrome

Heike Kölbel[1], *Kiran Polavarapu, Andreas Hentschel, Ozge Aksel Kilicarslan, Marc Pauper, Rachel Thompson, Ulrike Schara-Schmidt, Johann Böhm, Hanns Lochmüller, Andreas Roos*
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1.024P Muscular ciliopathy: a novel OCRL1 p.His524del variant is associated with skeletal muscle impairment in Lowe syndrome

Lola Albertella[1], *Ana Šarić, Baptiste Périou, Olivier Conscience, Marlène Rio, Cyril Gytiaux, Edoardo Malfatti, François Jérôme Authier, Ludovic Martin*
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1.025P Motor neuron vulnerability in triple-a syndrome: evidence from the largest genetically confirmed cohort

Menekse Oeztuerk[1], *Katja Dumić Kubat, Alexander Mensch, Daniel Natera de Benito, Maja Cehic, Sarah Leonard-Louis, Anna Codina, David Muhmann, Vera Dobelmann, Enrico Fruth, Anne Schänzer, Atchayaram Nalini, Vykuntaraju Kammasandra Nanjundagowda, Joachim Weis, Ulrike Schara-Schmidt, Johann Böhm, Stojan Peric, Andreas Roos, Tobias Ruck*
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1.026P A rare disease observed in the muscular dystrophy clinic: s-adenosylhomocysteine hydrolase (AHCY) deficiency

Hüseyin Per[1], *Dilek Türkmen, Sebahattin Çırak, Olcay Güngör, Uluç Yiş*
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Poster Session 1: 1.037eP- 1.054eP, 1.027P- 1.033P: Neurogenic Disorders - Clinical care and case reports

1.037eP Standing balance improvements with additional sensory input in Charcot-Marie-Tooth

Tarun Arora[1], *Fredrik Stenseth, Andreas Rosenberger, Kristian B. Nilsen, Christian Tronstad, Kristin Ørstavik*
[1]*Oslo University Hospital, Oslo, Norway*

1.038eP The progression of ankle range of motion in paediatric Charcot Marie Tooth (CMT): implications for function and strength

Evelin Milev[1], *Amy Wolfe, Emer O'Reilly, Charlotte Williams, Giovanni Baranello, Pinki Munot, Mary M Reilly, Francesco Muntoni*
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1.039eP Targeted search for the second variant in patients with SORD-neuropathy

Aysylu Murtazina[1], *Dmitrii Subbotin, Nikolay Lebedev, Dmitrii Pustoshilov, Nikita Bezkorovainyi, Oxana Ryzhkova, Artem Borovikov*
[1]*Research Centre for Medical Genetics, Moscow, Russia*

1.040eP Alpha-synuclein expression in skeletal muscle of amyotrophic lateral sclerosis patients: an immunohistochemical study

Can Ebru Kurt[1], *Zeynep Ergül-Ulger, Doruk Arslan, Sevim Erdem-Ozdamar, Ersin Tan*
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1.041eP ALS2-related pediatric motor neuron disease in six children

Serkan Kirik[1], *Onur Recep Gündüz, Ömer Bektaş*
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1.042eP A pilot study on AI vs. physician-led counseling for newly diagnosed ALS and GBS patients

Yigit Can Guldiken[1], *Seref Furkan Durgut*
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1.043eP Childhood chronic inflammatory demyelinating polyneuropathy: a single-center case series

Lutfiye Cilkol Bayram, ***Didem Ardıçlı***[1], *Hatice Bektas, Nesibe Gevher Eroglu Ertugrul, Gonul Vural, Aysegul Nese Citak Kurt*
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1.044eP Rethinking relapse detection in neuropathy: integrating patient-reported outcomes and objective functional tests

Camilla Hagedorn Thomsen[1], *Britt Stævnsbo Pedersen, Estephani Birkenfeldt Zeballos Medeiros, Sonja Desirée Holm-Yildiz, Jeppe Moesgaard Rasmussen, Tina Dysgaard*
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1.045eP Retrospective evaluation of pediatric Guillain–Barré syndrome according to 2023 European academy of neurology/peripheral nerve society guidelines

Ayşenur Çelik, ***Deniz Yüksel***[1], *Emrah Gün, Berna Uçan, Ece Akyüz, Bahadır Konuşkan*
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1.046eP Enhancing gait mechanics in paediatric Charcot Marie Tooth with lightweight carbon fiber ankle foot orthosis

Mahima Paul[1], *Vinitha Varghese, Immanuel Rajadurai Rajadurai, Sneha B.S. Shyamprasad, Himashweta Balwani, Sharanya S.P. Ravindra, Ann Agnes Mathew*
[1]*Bangalore Baptist Hospital, Bengaluru, India*

1.047eP RCC1-related acute-onset axonal neuropathy mimicking Guillain-Barré syndrome in two infants

Deniz Yüksel[1], *Nilay Ulukapı, Rojan İpek, Hamza Polat, Ali Cansu, Ayberk Türkyılmaz, İsmail Solmaz, Elifcan aşdelen, Bahadır Konuşkan, Haluk Topaloglu*
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1.048eP Clinical and genetic characteristics of pediatric hereditary neuropathies: a single-center experience

Deniz Yüksel[1], *Pınar Saka Ümit, Canan Üstün, Elifcan Taşdelen*
[1]*University of Health Sciences Turkey, Ankara Etlik City Hospital, Department of Pediatric Neurology and Neuromuscular Diseases Unit, Ankara, Turkey*

1.049eP Diagnostic course of chronic inflammatory demyelinating polyneuropathy spectrum with distal predominant weakness in Indonesia

Abraham Al Jody[1,2], *Muhammad Faruq Abdan Syakuron, Ahmad Yanuar Safri, Dinda Larastika Riyanto, Adrian Ridski Harsono, Nurul Fadli, Winnugroho Wiratman, Astri Budikayanti, Fitri Octaviana, Manfaluthy Hakim, Luh Ari Indrawati*
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1.050eP Evaluating muscle changes in persons with Charcot Marie Tooth disease using electrical impedance myography

Malin Eriksen[1], *Fredrik Stenseth, Carlos Robles, Kristian B. Nilsen, Seward Rutkove, Christian Tronstad, Ørjan G. Martinsen, Kristin Ørstavik, Tarun Arora*
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1.051eP Creatine kinase elevation mimicking myopathy in amyotrophic lateral sclerosis: a case of clinical diagnostic challenge

Sookin Moon[1], *Hyunjin Ju*
[1]*Nowon Eulji Medical Center, Seoul, South Korea*

1.052eP C9orf72 hexanucleotide repeat expansion presenting as progressive muscular atrophy with frontotemporal dementia
Yigit Can Guldiken[1], *Ebru Cesmeci, Mustafa Yagiz Ergin*
[1]*Canakkale Onsekiz Mart Universitesi, Canakkale, Turkey*

1.053eP A case atypical Guillain-Barre syndrome
Mustafa Cam[1], *Yiğit Can Güldiken*
[1]*Comu Tip Fakultesi Hastanesi, canakkale, Turkey*

1.054eP Advanced age Guillain Barre syndrome case
Mustafa Cam[1], *Yiğit Can Güldiken*
[1]*Comu Tip Fakultesi Hastanesi, canakkale, Turkey*

1.027P Co-designing a community-based physical activity program for children and adolescents with Charcot-Marie-Tooth disease
Rachel Kennedy[1,2], *Marlena Klaic, Evelyn Sloan, Archita Joshi, Bridget Taylor, Kate Carroll, Peter Critchley, Eppie Yiu*
[1]*Murdoch Children's Research Institute, Parkville, Australia,* [2]*Royal Children's Hospital, Parkville, Australia*

1.028P Baseline glycolipid IgG autoantibodies in GBS are prevalent and show cross-regional pattern concordance
Anton Nguyen[1], *Preeti Paliwal, Susan Halstead, Henk-André Kroon, David Lorenz, Zhahirul Islam, Kazim Sheikh, Kenneth C. Gorson, Hugh Willison*
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1.029P Expanding phenotypic spectrum of SORD related Charcot-Marie-Tooth disease: a study from India
Dipti Baskar[1], *Atchayaram Nalini, Seena Vengalil, Saraswati Nashi, Gautham Arunachal, Ananthapadmanabha Kotambail*
[1]*National Institute of Mental Health and Neuro Sciences (NIMHANS), Bengaluru, India, Bengaluru, India*

1.030P Determinants of comorbidities influencing quality of life in pediatric neuropathy patients: implications for optimized follow-up strategies
Gizem Aktemur, ***Gulten Ozturk Thomas***[1], *Olcay Unver, Hakki Akbeyaz, Dilsad Turkdogan*
[1]*Marmara University Pendik Education and Research Hospital, Istanbul, Turkey*

1.031P Inherited neuropathies in Senegal: clinical, electrophysiological and genetic features from the Centre Hospitalier National Universitaire (CHNU) de Fann, Dakar
Henriette Viéra Frichérie Senghor Dite Aida[1], *Ayoub Mouhamed Saleh, Maouly Fall, Marième Soda Diop-Sène, Anna Modji Basse Faye, Adjaratou Djeynaba Sow, Moustapha Ndiaye, Mamadou Sy, Lala Bouna Seck, Ngor Side Diagne, Pedro M Rodriguez Cruz*
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1.032P Genetic epidemiology of adult-onset hereditary neuropathies in Serbia
Milica Vukojevic[1], *Ana Marjanovic, Vukan Ivanovic, Jovan Pesovic, Ana Kosac, Ayse Candayan, Milena Jankovic, Dusanka Savic-Pavicevic, Albena Jordanova, Ivana Basta, Stojan Peric*
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1.033P Defining the clinical, neurophysiological, and neuroimaging features of recently described PIGG-associated motor neuropathy
Anastasia Vlasenko[1], *Dmitrii Subbotin, Andrey Marakhonov, Artem Borovikov, Aysylu Murtazina*
[1]*Research Centre for Medical Genetics, Moscow, Russia*

Poster Session 1: 1.055eP- 1.058eP, 1.034P- 1.036P: Neurogenic disorders - Imaging and outcome measures

1.055eP Quantitative ALS: development of a novel, physician-reported, ALS outcome measure
Pola Gaid[1], *Namita A Goyal*
[1]*University of California, Irvine, Irvine, CA, United States*

1.056eP Muscle ultrasound feature-based predictive modeling for amyotrophic lateral sclerosis staging: machine learning approaches
Joo Hye Sung[1], *Young-Bin Park, Jin-woo Park, Byung-Jo Kim*
[1]*Chuncheon Sacred Heart Hospital, Chuncheon, South Korea*

1.057eP Continuous real-world assessment of ambulation in amyotrophic lateral sclerosis using digital health technology

Camila Gonzalez Barral[1], *Margaux Poleur, Katherine M. Burke, Oihana Piquet, Charlotte Rasser, Georgia Stimpson, Kendall Carney, Sydney Hall, Sravan Mandepudi, Timothy Royse, Narghes Calcagno, Charlotte Hermanne, Stéphanie Delstanche, Damien Eggenspieler, James D. Berry, Laurent Servais*
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1.058eP Accelerating ALS trials through muscle-centered assessment

Jean-yves Hogrel[1], *Ghida Trad, Timothée Lenglet, Isabelle Ledoux, Giorgia Querin, Véronique Marchand-Pauvert, Pierre-François Pradat*
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1.034P Shared and distinct suprahyoid ultrasonographic patterns in ALS and neuromuscular dysphagia: implications for subclinical detection

Young Bin Park[1], *Joo Hye Sung, Jin Woo Park, Byung Jo Kim*
[1]*Korea University Anam Hospital, Seoul, South Korea*

1.036P A novel Guillain–Barré syndrome clinical progression scale to characterise the patient's care journey: utilisation in a phase 3 trial of tanruprubart

Anton Nguyen[1], *Henk-André Kroon, Kenneth C. Gorson, Quazi Deen Mohammad, Thomas Harbo*
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Poster Session 1: 1.037P-1.038P: Neurogenic disorders – Physiopathology

1.037P Differential regulation of MuRF1 expression in amyotrophic lateral sclerosis and disuse-induced muscle atrophy in mice

Tsukasa Tominari[1], *Masaru Takatoya, Masaki Inada, Yoshitsugu Aoki*
[1]*Department of Molecular Therapy, National Center of Neurology and Psychiatry, Tokyo, Japan,*

1.038P From skeletal muscle regeneration and aging to ALS ... and back again

Sonia Alonso Martin[1,2], *Oihane Pikatza-Menoio, Amaia Elicegui, Sandra Fuertes-Alvarez, Maria Levchuk, Mikel Garcia-Puga, Ainhoa Vidal-Gil, Nora Hernandez-Montalvo, Adolfo Lopez de Munain, Haser H. Sutcu*
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Poster Session 1: 1.039P-1.043P: Neurogenic Disorders – Treatments

1.039P Long-term evaluation of tofersen in SOD1-ALS: final integrated results from the phase 1/2 ascending dose trial and the open-label extension

Stephanie Fradette[1], *Timothy Miller, Robert Bucelli, Merit Cudkowicz, Pamela Shaw, Manjit McNeill, Kaelin O'Connell, Jennifer Inra, Steve Garafalo*
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1.040P An open-label study evaluating tanruprubart in participants from North America and Europe with Guillain-Barré syndrome

Rafid Mustafa, Shafeeq Ladha, Kavita Grover, Glenn Morrison, Ping Lin, ***Anton Nguyen***[1], *Henk-André Kroon, Jeffrey A. Allen*
[1]*Annexon Biosciences, Brisbane, United States, 5University of Minnesota, Minneapolis, United States*

1.041P Tanruprubart in GBS: targeting early complement-associated inflammation conserved across the clinical spectrum and regions

Anton Nguyen[1], *Kazim Sheikh, Preeti Paliwal, Henk-André Kroon, Zhahirul Islam, David Lorenz, Edmund Chang, Yuanbin Ru, Ted Yednock, Rick Artis, Claudia Sommer*
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1.042P RNA aptamers that target glutamate receptors are efficacious in rescuing disease phenotypes in both sporadic and familial ALS mouse models

Li Niu[1], *Megumi Akamatsu*
[1]*University at Albany, State University of New York, Albany, United States*

1.043P Nonclinical efficacy and safety evaluation of SGT-212, a novel investigational AAV FXN gene therapy for the treatment of Friedreich's ataxia

Brandon YH Chan, Jun Lee, Jamie Marshall, ***Matthew Harmelink***[1], *Gourav Roy Choudhury, Heather Born, Juliette Hordeaux, James M Wilson, Jessie Hanrahan, Gabriel Brooks, Nicolas Christoforou*
[1]*Solid Biosciences, Charlestown, United States*

Poster Session 1: 1.059eP, 1.044P-1.050P: Novel molecular genetic approaches for NMD

1.059eP Application of nanopore sequencing for the diagnosis of muscle disorders

Milla Laarne[1,2], *Teemu Turku, Harri Kangas, Kati Donner, Taina Turunen, Vilma-Lotta Lehtokari, Katarina Pelin*
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1.044P Clinical and genetic features of childhood-onset hereditary spastic paraplegias: a multicenter study

Akgun Olmez, Mustafa Komur, Ezgi Caglar, Burcin Gonullu Polat, Semra Hiz, Uluc Yis, Ayse ***Ipek Polat***[1], *Yavuz Oktay, Hasan Tekgul, Sanem Keskin Yilmaz, Erdem Simsek, Didem Ardicli, Ilknur Erol, Seyda Besen, Ulviyya Guliyeva, Deniz Yuksel, Gokhan Ozan Cetin, Kadir Intas, Haluk Aydin Topaloğlu*
[1]*Dokuz Eylül University, Faculty of Medicine, Department of Pediatrics, Division of Pediatric Neurology, izmir, Turkey*

1.045P Exploring the molecular landscape of Friedreich ataxia with long-read sequencing

Sebastien Audet[1], *Marjorie Labrecque, Valerie Triassi, Carolane Charest, Adrien Rihoux, Laura Hamilton, Grace Yoon, Antoine Duquette, Martine Tetreault*
[1]*CHUM Research Center, Montreal, Canada*

1.046P MyoSpliceAtlas: alternative splicing landscapes of cardiac and skeletal muscle reveal mechanisms of development and disease

Ali Oghabian[1], *Per Harald Jonson, David Gomez Andres, Francina Munell, Jessica Camacho Soriano, Maria Angeles Sanchez Duran, Juha Sinisalo, Heli Tolppanen, Johanna Tolva, Peter Hackman, Bjarne Udd, Marco Savarese*
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1.047P Evidence suggesting a common protein aggregation mechanism across missense variants in actin-binding domains of neuromuscular disease genes

Johanna Ranta-aho[1,2], *Per Harald Jonson, Bjarne Udd, Marco Savarese*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Helsinki, Finland*

1.048P MAMDC2 mutations dysregulate human muscle cells proliferation and differentiation

Emilia Servian Morilla[1], *Victoria Navarro Garrido, Maria Isabel Ibañez Cerezuela, Pedro Javier Gómez Galvez, Cristina Domínguez González, Ana Rita Gonçalves, Celia Azevedo Soares, Rosário Santos, Henrique Moniz Costa, Gina Ravenscroft, Carmen Paradas López, Macarena Cabrera Serrano*
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1.049P mRNA-LNP myostatin propeptide therapy promotes age-dependent skeletal muscle hypertrophy in mice

Brock Wetzlich[1], *Jinzeng Yang*
[1]*University Of Hawaii At Manoa, Honolulu, United States*

1.050P Improved detection of long neuromuscular disease transcripts with ribosomal RNA depletion RNA-Seq

Swethaa Natraj Gayathri[1,2], *Victoria Lillback, Bjarne Udd, Peter Hackman, Marco Savarese, Ali Oghabian*
[1]*University of Helsinki, Helsinki, Finland,* [2]*Folkhälsan Research Center, Helsinki, Finland*

Poster Session 1: 1.060eP- 1.088eP, 1.051P-1.073P: SMA - Clinical care and case reports

1.060eP Healthcare utilization and associated barriers among adults living with spinal muscular atrophy in the United States

Erin Welsh[1], *Sarah Whitmire, Mary Curry, Lisa Belter*
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1.061eP Real-world treatment with risdiplam in adults with spinal muscular atrophy (SMA): a single center experience

Hacer Durmus[1], *Sezan Mergen, Arman Cakar, Yesim Parman*
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1.062eP Advancing care in spinal muscular atrophy through patient-centred, motor-focused rehabilitation: prospective interventional outcomes from the ACE SMA study

Charlotte Lilien[1], *Foteini Mavrommati, Serena Hill, Georgia Stimpson, Hayley Ramjattan, Francesca Taylor, Johnny Collett, Laurent Servais, SMA REACH UK Network*
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1.063eP Priorities in information sharing and management in spinal muscular atrophy (PRISM-SMA), a mixed methods study

William Quelch[1], *Portia Thorman, Ulrika Kreicbergs, Min Ong*
[1]*Leeds Teaching Hospital NHS Trust, Leeds, United Kingdom*

1.064eP The natural history and treatment effects of SMA patients with ≥4 SMN2 copies in the United States

Chamindra Laverty[1], *Randell Richardson, Kuntz Nancy, Kaitlin Batley, Susan Apkon, Julie Parsons, Diana Bharucha-Goebel, Sarah Wright, Katherine Mathews, Dimah Saade, Leigh Ramos-Platt, Julian Thomas, Emma Ciafaloni, Cuixia Tian, Omer Abdulhamid, Kapil Arya, Bakri Elsheikh, John Brandsema, Russell Butterfield, Carolina Tesi-Rocha*
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1.065eP Impact of early treatment initiation on ambulatory outcomes in SMA type 3a: insights from Spanish and Italian real-world cohorts

Andres Eduardo Nascimento Osorio[1], *Rocio Garcia-Uzquiano, Cristina Puig-Ram, Giorgia Coratti, Mireia Alvarez Molinero, Karolina Aragon-Gawinska, Chiara Bravetti, Chiara Cutrì, Adele D'Amico, Miguel Angel Fernandez Garcia, Stephanie Lotz Esquivel, Riccardo Masson, Sonia Messina, Carolina Ñungo Garzon, Marika Pane, Carmen Paradas, Elena Pegoraro, Valeria Sansone, Eugenio Mercuri, .CUIDAME and ITASMAC Investigators Group*
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1.066eP Case Report: Phenotypic diversity in a family with SMN1 homozygous deletion and 4 copies of SMN2

Esra Sarigecili[1], *Ahu Ekren, Habibe Koc Ucar, Tamer Celik, Yilmaz Akbas, Mustafa Kömür*
[1]*Adana Sehir Hastanesi, Çocuk Nöroloji, Adana, Turkey*

1.067eP Evaluation of motor function in patients with SMA types 2 and 3 receiving disease-modifying therapy: a single-center experience

Sema Saltık[1], *Yasemin Sancak, Seda Sönmez Kurukaya, Neslihan Özhan, Esma Şengenç*
[1]*Istanbul University-Cerrahpaşa, Cerrahpaşa Faculty of Medicine, Department of Pediatrics, Division of Pediatric Neurology, Istanbul, Turkey*

1.068eP Prospective study protocol investigating nutrition, bulbar and motor function in infants with spinal muscular atrophy

Katie Obrien[1,2,3], *Kay Nguo, Eppie Yiu, Ian Woodcock, Kate Carroll, Angela Radatti-Brown, Daniella Villano, Natassja Billich, Zoe Davidson*
[1]*Monash University, Melbourne, Australia,* [2]*Royal Children's Hospital, Melbourne, Australia,* [3]*Murdoch Children's Research Institute , Melbourne, Australia*

1.069eP Beyond the floor effect: long-term respiratory stabilization with risdiplam in severe adult SMA type 2, a single centre experience

Sara Loprieno[1], *Francesca Torri, Marco Gherardi, Massimiliano Serradori, Laura Carrozzi, Gabriele Siciliano, Giulia Ricci*
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1.070eP The rarest of the rare: a case series of spinal muscular atrophy with lower extremity predominance type 1 (SMA-LED1)

Ayse Nur Ozdag Acarli[1], *Ayfer Arduc Akcay, Ilaha Bakirli, Sahin Avci, Hulya Kayserili, Piraye Oflazer*
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1.071eP Clinical characteristics and treatment outcomes in patients with spinal muscular atrophy and four SMN2 copies

Anna Lemska[1], *Sylwia Studzińska, Maria Mazurkiewicz-Beldzinska*
[1]*Medical University of Gdańsk, Gdansk, Poland*

1.072eP SMA Care UK: optimising bone health for individuals with SMA across the UK

Vanessa Christie-Brown[1,2], *James B. Lilleker, Stephanie A. Borg, Judith Bubbear, Alexander D. Chesover, Emma M. Clark, Nicola Crabtree, Christian de Goede, Emma L. Duncan, Lydia Forestier-Zhang, Channa Hewamadduma, Peter Selby, Sze Choong Wong, Claire Wood, Chiara Marini-Bettolo, Anne-Marie Childs*
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1.073eP SMA Care UK: management of hips in SMA in the DMT era - UK multidisciplinary consensus recommendations

Vanessa Christie-Brown[1,2], *Emer O'Reilly, Maria I. Vanegas, Jennie Sheehan Sheehan, Caroline Batke, Channa Hewamadduma, Iain Horrocks, Claire Murnaghan, Lindsey Pallant, Rosanna Rabb, Anne-Marie Childs, Chiara Marini-Bettolo, Fabian Norman-Taylor, Michail Kokkinakis, Giovanni Baranello, On behalf of the SMA Care UK Hip Working Group*
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1.074eP Rehabilitation needs and care models in spinal muscular atrophy: a real-world analysis from an italian community rehabilitation center

Anna Lia Frongia[1,2], *Marta Briganti, Anna Lisa Branca, Valentina Saraceni, Eleonora Torchia, Salvatore Rossi, Paola Orlando, Nicoletta Mandis, Rebecca Della Rocca, Mauro Onofri, Ornella Robertazzi, Cristina Sancricca*
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1.075eP Developing exercise guidelines for spinal muscular atrophy: an SMA care UK initiative

Charlotte Lilien[1], *Robert Muni-Lofra, Peter Waldock, Vanessa Christie Brown, Chiara Marini-Bettolo, Anne-Marie Childs, . on Behalf of the SMA CARE UK- Exercise guidelines working group*
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1.076eP Real-world outcomes of early gene therapy for SMA following newborn screening

Tanitnun Paprad[1,2], *Hernan Gonorazky, Elisa Nigro, Lauren Weinstock*
[1]*The Hospital For Sick Children, Canada, Toronto, Ontario, Canada,* [2]*King Chulalongkorn Memorial Hospital, Bangkok, Thailand*

1.077eP Nationwide newborn screening for spinal muscular atrophy during active military conflict: the Ukrainian implementation experience

Svitlana Kormoz[1], *Nataliia Olkhovych, Nataliia Mytsyk, Oksana Barvinska, Nataliia Samonenko, Tetiana Shkliarska, Nataliia Gorovenko*
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1.078eP Adult SMA REACH: characterisation of adult patients living with SMA at start of treatment

Julie Bohill[1], *Elena Karkkainen, Laura Simms, Jose Verdu-Diaz, Aleks Carver, Stephanie Tanner, Jassi Michell-Sodhi, Dionne Moat, Robert Muni Lofra, Chiara Marini-Bettolo*
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1.079eP The use of spinal bracing in severe spinal muscular atrophy: a multicentre audit

William Quelch[1], *Kay White, Steve Hewitt, Francesca Henderson, Emer O'Reilly, Jose Longatto, Chiara Brusa, Alison Skippen, Sithara Ramdas, Mariacristina Scoto, Shreya Srinivas, Min Ong*
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1.080eP Adult SMA REACH: a comparative UK real world data study to assess the safety and efficacy profiles of nusinersen and risdiplam in adults with SMA

Julie Bohill[1], *Elena Karkkainen, Laura Simms, Jose Verdu-Diaz, Aleks Carver, Grecia Alvarez, Stephanie Tanner, Dionne Moat, Robert Muni Lofra, Chiara Marini Bettolo*
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1.081eP UK real-world data collection study to analyse respiratory trajectories in SMA patients undergoing treatment with nusinersen and risdiplam

Julie Bohill[1], *Robert Muni Lofra, Laura Simms, Jose Verdu-Diaz, Aleks Carver, Stephanie Tanner, Jassi Michell-Sodhi, Dionne Moat, Elena Karkkainen, Chiara Marini Bettolo*
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1.082eP Motor trajectories after risdiplam following gene therapy in SMA type 1

Revital Lavi[1,2], *Anna Shtamler, Sharona Katzenellenbogen, Hagar Tebul, Anat Weizman, Liora Sagi*
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1.083eP Motor outcomes of nusinersen therapy in spinal muscular atrophy type 2: analysis of the national cohort from Kazakhstan

Begimay Zholdasbekova[1], *Latina Tekebayeva*
[1]*University Medical Center, Astana, Kazakhstan*

1.084eP Changes in functional mobility following generic risdiplam spinal muscular atrophy: a 6-month observational study

Gauri Khandekar[1], *S.B. Shyamprasad, S.P Ravindra, Himshweta Balwani, Ann Agnes Mathew*
[1]*Synapse Neuro and Child Development Center, Bengaluru, India,* [2]*Virgo healthcare center, Bengaluru, India*

1.085eP Phenotypic discordance and near-presymptomatic gene therapy in dizygotic twins with SMA type 1: evidence for modifier effects beyond SMN2 copy number

Yun Jeong Lee[1], *Young Tae Lim, Ha Neul Lee*
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1.086eP Beyond the classical phenotype: a homozygous ASAH1 variant presenting with juvenile-onset myoclonic epilepsy

Deniz Yüksel[1], *Hilal Akalın, Ayşe Bayram*
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1.087eP When spinal muscular atrophy is there but the test says no: a case of somatic mosaicism leading to false-negative diagnosis
Milos Brkusanin[1], *Slobodanka Todorovic, Dragana Vucinic, Dusanka Savic-Pavicevic*
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1.088eP "Double-trouble" with spinal muscular atrophy: diagnostic complexity and therapeutic implications
Phisitphon Ounchokdee[1,2], *Tanitnun Paprad, Freddy Paiz, Emily Povse, Nicole Park, Hernan Gonorazky, Grace Yoon*
[1]*The Hospital for Sick Children, University of Toronto, Toronto, Canada,* [2]*Ramathibodi Hospital, Mahidol University, Bangkok, Thailand*

1.051P Orthotic management in spinal muscular atrophy (SMA) in the era of disease-modifying therapies
Olivia Martineau[1], *Fabian Norman-Taylor Norman-Taylor, Adnan Manzur, Francesco Muntoni, Lianne Abbott, Rachel Thomas, Jose Longatto, Rafael Terepolsky, Giovanni Baranello, Mariacristina Scoto*
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1.052P Gait analysis in new cohort of paediatric ambulant spinal muscular atrophy type 1 and type 2 patients, and the role of orthotics
Alexander Schofield[1], *Jennie Sheehan, Caroline Smith, Maria Vanegas*
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1.053P A rare DYNC1H1-related neuro-orthopedic phenotype with SMA, arthrogryposis and congenital fractures: a case series
Evgeniya Melnik[1], *Vladimir Kenis, Kseniya Zabudskaya, Vadim Tsargush, Sergei Nikitin, Dmitrii Buklaev, Elena Dadali*
[1]*Research Center for Medical Genetics, Moscow, Russia*

1.054P Longitudinal serum neurofilament light trajectories following treatment initiation or modification in spinal muscular atrophy
Gianpaolo Cicala[1,2], *Anna Capasso, Fiorella Piemonte, Sara Petrillo, Maddalena Bianchetti, Giorgia Coratti, Romina Venditti, Martina D'Ambrosio, Laura Castellini, Martina Malaspina, Adele D'Amico, Sonia Messina, Valeria Sansone, Stefania Corti, Marika Pane, Danilo Francesco Tiziano, Eugenio Mercuri*
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1.055P Multi-domain response analysis of adult spinal muscular atrophy patients treated with risdiplam: insights from a real-world two-center French study
Claire Lefeuvre, Jean-Yves Hogrel, Frederic Fer, Marion Masingue, Marjorie Banos, Edouard Berling, Anthony Behin, Sarah Leonard-Louis, Guillaume Bassez, Gorka Fernandez, Alice Rouyer, Clement Guemy, Noemie Petit, Fanny Chantreuil, Sylvain Petit, Guillaume Nicolas, Pascal Laforet, Tanya Stojkovic, ***Tanya Stojkovic***[1]
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1.056P Timing of therapy in sma type 1: longitudinal clinical and real-world insights
Hakkı Akbeyaz, Gülten Öztürk, Olcay Uner, Evrim karadag, ***Dilsad Turkdogan***[1]
[1]*Marmara Medical Faculty, Department of Pediatric Neurology, Istanbul, Turkey*

1.057P Neurodevelopmental outcomes and determinants in spinal muscular atrophy across the newborn screening era
Didem Ardıçlı[1], *Pelin Celik, Funda Akpinar, Nesibe Gevher Eroglu Ertugrul, Aysegul Nese Citak Kurt*
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1.058P Shifting referral paradigms following introduction of generic risdiplam in India: a single centre experience
Himshweta Balwani[1,2], *Sneha Shyamprasad, Gauri Khandekar, Sharanya Ravindra, Ann Mathew*
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1.059P Association between cerebrospinal fluid neurofilament light chain levels and motor function in spinal muscular atrophy patients with 3-4 SMN2 copies
Chen-hua Wang[1,2,3], *Wen-Chen Liang, Hui-Min Hsieh, Yuh-Jyh Jong*
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1.060P SMA Care UK: ensuring the best management of spine for individuals with SMA across the UK

Vanessa Christie-Brown[1,2], *Lianne Abbott, Andrew James Bowey, Sherryl Chatfield, Sarah Gregson, Steven Hewitt, Charlotte Lillien, Emma Manchester, Eileen Morrow, Katie Nevin, Min T. Ong, Emer O'Reilly, Simon Roberts, Mariacristina Scoto, Jennie Sheehan, Athanasios Tsirikos, Chiara Marini-Bettolo, Anne-Marie Childs, Robert Muni-Lofra, On behalf of the SMA Care UK Spine Working Group*
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1.061P SMA Care UK: ensuring the best respiratory care for individuals with SMA

Vanessa Christie-Brown[1,2], *Ben Messer, Elaine Chan, Jennifer Carter, Lisa Edel, Christopher Edwards, Marjorie Illingworth, Patrick Murphy, Tracy Procter, Martin Samuels, Portia Thorman, Chiara Marini-Bettolo, Anne-Marie Childs*
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1.062P SMA Care UK: a national initiative to ensure that those living with SMA in the UK receive the best possible care

Vanessa Christie-Brown[1,2], *Giovanni Baranello, Grecia Benesperi, Claire Fitzpatrick, Channa Hewamadduma, Iain Horrocks, James Lilleker, Giles Lomax, Grace Marston, Robert Muni-Lofra, Neeru Naik, Emer O'Reilly, Mariacristina Scoto, Portia Thorman, Andi Thornton, Chiara Marini-Bettolo, Anne-Marie Childs*
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1.063P SMA Care UK: ensuring high-quality transition from paediatric to adult care in SMA, in the era of disease-modifying therapies

Vanessa Christie-Brown[1,2], *Lisa Cutsey, Becci Howell, Emma Manchester, Olivia Martineau, Grace Mc Maken, Michele Phillips, Andy Rose, Victoria Selby, Natalie Smith, Rachel Thomas, Tracey Willis, Denise Wooding, Anne-Marie Childs, Chiara Marini-Bettolo*
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1.064P Adrenal and bone complications in paediatric patients living with spinal muscular atrophy – a single centre retrospective study

Patricia Prianca Ramji[1], *Lydia Forestier-Zhang, Rosa Boursin, Elspeth C Ferguson, Stephanie Borg, Charlotte J Elder, Min Tsui Ong*
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1.065P Pregnancy outcomes and experiences in women with spinal muscular atrophy in the era of disease-modifying therapies: an international study

Simone Thiele[1], *Miriam Hiebeler, Marcel Heidemann, Eva Malm, Nicolas Schiermeier, Veronika Bittner, Aleks Carver, Anna Łusakowska, Lenka Mokrá, Miguel Oliveira Santos, Olesja Parmova, Miriam Rodrigues, Victoria Hodgkinson, Hanns Lochmueller, SMArtCARE study group and steering committee, Maggie C. Walter*
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1.066P Newborn screening for spinal muscular atrophy using a multiplex molecular approach: five-year results from a regional program

Concetta Aloi, Monica Traverso, Alessandro Salina, Michela Cassanello, Barbara Tappino, Marina Pedemonte, Noemi Brolatti, Federica Lanza, Stefano Volpi, Federico Zara, Mohamad Maghnie, ***Claudio Bruno***[1,2]
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1.067P Long-term real-world clinical and functional outcomes in adults with 5q spinal muscular atrophy treated with nusinersen

Veronica Ivonne Lopez Diaz[1], *Maria Elena Meza-Cano*
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1.068P Ultra rapid treatment in newborns with spinal muscular atrophy (SMA) with 2 SMN2 copies: a multicenter, retrospective cohort study

Natalie Goedeker[1], *Kapil Arya, Kaitlin Batley, John Brandsema, Michelle Farrar, Marcia Felker, Mark Fisher, Melissa Gibbons, Karen Herbert, Nancy Kuntz, Regina Laine, Lakhotia Arpita, Bohoon Lee, Omer Abdul Hamid, Seth Perlman, Randal Richardson, Amanda Rogers, Aravindhan Veerapandiyan, Zurisadai Gonzalez Castillo, Craig Zaidman*
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1.069P Functional stability in adult SMA patients treated with risdiplam: a real-world data

Eshgin Maharramov, Doruk Arslan, Fatma Ayvat, Muhammed Kılınç, Sevim Erdem-Ozdamar, Ersin Tan, Can ***Ebru Kurt***[1]
[1]Hacettepe University, School of Medicine Department of Neurology, Ankara, Turkey

1.070P Early real-world adoption of high-dose nusinersen in patients with spinal muscular atrophy: patient preference and initial clinical observations from Japan

Hideyuki Iwayama[1], *Shingo Numoto, Yoshiteru Azuma, Hirokazu Kurahashi, Yumiko Yasue, Hiroyuki Kawajiri, Atsushi Yanase, Teruyoshi Ito, Takahiro Ogawa, Yoshinori Ito, Akihisa Okumura*
[1]*Aichi Medical University, Nagakute, Japan*

1.071P Real-world clinical outcomes of onasemnogene abeparvovec in Korean patients with spinal muscular atrophy: a 36-month retrospective nationwide cohort study

Seungbok Lee[1], *Jihye Kim, Noorhee Son, Byung Jin Lee, Ji-Hye Byun, Ra Won Kang, Yun Jeong Lee, Anna Cho, Byung Chan Lim, Soyoung Yi, Jong-Hee Chae*
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1.072P Real-world 12-month outcomes of risdiplam in spinal muscular atrophy types 2 and 3: a Brazilian cohort

Erick Dupont[1,2], *Clara Camelo, Rodrigo Mendonça, Cristiane Moreno, Graziela Polido, Eduardo Vital, Weverton Teixeira, Daniel Hayashi, Joemir Brito, Ciro Júnior, Edmar Zanoteli*
[1]*Universidade de São Paulo - USP, São Paulo, Brazil,* [2]*Hospital das Clínicas da Faculdade de Medicina de Universidade de São Paulo - HCFMUSP, São Paulo, Brazil*

1.073P Four-year trajectory of children with SMA receiving nusinersen in the United Kingdom

Emer O'reilly[1], *Deborah Ridout, Georgia Stimpson, Nikki Cornell, Jennie Sheehan, Maria Vanegas, Elizabeth Wraige, Lindsey Pallant, Anne-Marie Childs, Sarah D'Urso, Min Ong, Marion Main, Lianne Abbott, Chiara Marini-Bettolo, Pinki Munot, Adnan Manzur, Giovanni Baranello, Francesco Muntoni, Mariacristina Scoto, SMA REACH UK Network*
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Poster Session 1: 1.089eP-1.097eP, 1.074P- 1.082P: SMA - Imaging and outcome measures

1.089eP Functional outcomes in infants with spinal muscular atrophy (SMA) type 1: motor, respiratory, and bulbar domains

Lisa Edel[1], *Deborah Ridout, Georgia Stimpson, Emily Johnson, Lianne Abbott, Olivia Martineau, Adnan Manzur, Mariacristina Scoto, Francesco Muntoni, Giovanni Baranello*
[1]*UCL GOS Institute of Child Health, London, United Kingdom*

1.090eP Facial nerve diameter as a potential biomarker in adults with spinal muscular atrophy

Felipe Franco Da Graca[1], *Guilherme Soares de Oliveira Wertheimer, Thiago Junqueira Ribeiro de Rezende, Cristina Iwabe, Rodrigo de Holanda Mendonça, Edmar Zanoteli, Marcondes Cavalvante França Jr*
[1]*University Of Campinas, Campinas, Brazil*

1.091eP Correlating EK2 and RULM to assess upper limb function in SMA

Emer O'Reilly[1], *Amy Wolfe, Georgia Stimpson, Giovanni Baranello, Francesco Muntoni, Mariacristina Scoto*
[1]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom*

1.092eP User experience in a home-based exergame in youth with spinal muscular atrophy: preliminary findings from a national clinical trial

Allison Grangé[1,2], *Ariane Belzile, Sarah Deckarm, David Herzig, Maryam Oskoui, Danielle Levac*
[1]*Université De Montréal, Montréal, Canada,* [2]*Azrieli Research Center UHC Ste Justine, Montréal, Canada*

1.093eP Evaluation of motor function in patients with spinal muscular atrophy using finger movements

Toshio Saito[1], *Hisahide Nishio, Kimimasa Tobita*
[1]*NHO Osaka Toneyama Medical Center, Toyonaka, Japan*

1.094eP Smartphone-based two- versus six-minute walk test for remote assessment of gait

Rachele Rossanigo, Wei Wei, Adrien Juraver, Corrado Bernasconi, Óscar Reyes, Shibeshish Belachew, ***Claudia Mazzà***[1]
[1]*Indivi AG, Basel, Switzerland*

1.095eP Translation and cross-cultural adaption of the functional ability scale for evolving spinal muscular atrophy (EVOLVE-SMA) into Spanish– escala de capacidad funcional para atrofia muscular espinal evolutiva

Meghan Moore Burk[1], *Magalí Squitin Tasende, Javier Muntadas, Javiera Ortega, Diana Rodriguez, Maria Abila, Amanda Appel*
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1.096eP Telemedicine enabled continuum of care in spinal muscular atrophy

Sneha Banavara Shyamprasad[1], *Gauri Khandekar, Himshwetha Balwani, Ann Agnes Mathew*
[1]*Synapse Neuro Center, Bengaluru, India*

1.097eP The integration of PROMs and real-world data: a holistic approach to characterise disease burden and treatment impact in spinal muscular atrophy

Julie Bohill[1], *Aleks Carver, Elena Karkkainen, Dionne Moat, Stephanie Tanner, Elzo Cavalcante, Kate Adcock, Maria Farrugia, Gennadiy Ilyashenko, James Lilleker, John McConville, Andria Merrison, Matt Parton, Liz Ryburn, Robert Munri Lofra, Portia Thorman, Mariacristina Scoto, Chiara Marini Bettolo*

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1.074P Evaluation of an artificial intelligence–based approach to disease severity estimation using stimulus-evoked muscle activity in patients with spinal muscular atrophy

Sooyeon Kim[1], *Jin Hong Shin, Kwangsub Song, Sangui Choi, Hooman Lee, Howon Jeon*

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1.075P Patterns of cardiopulmonary and muscle oxygenation response to assisted cycle (A6MCT) exercise in individuals with neuromuscular diseases

Tina Duong[1], *Audrey Chun, Constance de Monts, Melina Rogers, Sally Dunaway Young, Shelby Vogt-Domke, Raiye Hailu, Sarah Ismail, Lin Karman, Sabrina Salvatore, Whitney Tang, John Day, Tanja Taivassalo*

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1.076P Electrophysiological biomarkers in spinal muscular atrophy

Jesper Storgaard[1,2], *Erisela Qerama, Hatice Tankisi, Henning Andersen*

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1.077P Beyond the clinic: multiday analysis of leg movement quantity and kinematic characteristics in infants with SMA and infants with typical development

Melissa McIntyre[1], *Jinseok Oh, Tina Duong, Meghan Moore Burk, Sophia Olson, Michelle Loftus, Stephanie Manberg, Kristen Wong, Danica Reader, Amelia Wilson, Russell J Butterfield, Beth A Smith*

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1.078P Longitudinal evaluation of cervical spinal cord MRI in adult 5q-SMA: potential biomarkers and pathophysiological insights

Felipe Franco Da Graca[1], *Thiago Junqueira Ribeiro de Rezende, Cristina Iwabe, Carelis González Salazar, Rodrigo de Holanda Mendonça, Edmar Zanoteli, Marcondes Cavalcante França Jr.*

[1]*University Of Campinas, Campinas, Brazil*

1.079P Modeling treatment response trajectories in spinal muscular atrophy: results from the MAP THE SMA study

Giorgia Coratti[1], *Gaia Camisasca, Jacopo Lenkowicz, Carlotta Masciocchi, Chiara Bravetti, Stefano Patarnello, Marika Pane, Eugenio Mercuri, Paulo Gabriel Longo*

[1]*Catholic University of Sacred Heart, Rome, Italy*

1.080P Multivariable prediction of treatment response in type II SMA: beyond single outcome measures

Giorgia Coratti[1], *Francesca Bovis, Chiara Cutrì, Sophia Paolucci, Emilio Albamonte, Valeria Sansone, Adele D'amico, Sonia Messina, Claudio Bruno, Caterina Agosto, Tiziana Mongini, Federica Ricci, Massimiliano Filosto, Marika Pane, Eugenio Mercuri*

[1]*Catholic University of Sacred Heart, Rome, Italy*

1.081P Validation of a new multidimensional clinical outcome measure for evaluating adolescent and adult patients with spinal muscular atrophy

Carolina Ñungo Garzón, Esther Toro Tamargo, Mónica Povedano Panades, Miguel Ángel Fernández-García, Iñigo Rojas Marcos, Laura González Mera, Sofía García-López, Pablo Rebollo, Albert Rafels, Ángeles Terrancle, María Grazia Cattinarl, Mercedes Martínez-Moreno, Jorge Mauriño, ***Juan Francisco Vazquez Costa***[1,2,3]

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1.082P Patient-centered outcomes in adolescents and adults with spinal muscular atrophy treated with risdiplam

Juan Francisco Vazquez Costa[1], *Nancy Carolina Ñungo Garzón, Karolina Aragon-Gawinska, Inmaculada Pitarch Castellano*

[1]*University of Valencia, Valencia, Spain*

Poster Session 1: 1.098eP- 1.099eP, 1.083P- 1.087P: SMA – Physiopathology

1.098eP Investigation of long non-coding RNA profiles derived from peripheral blood mononuclear cells in spinal muscular atrophy

Ayse Ipek Polat[1], *Uluc Yis, Semra Hiz, Yavuz Oktay Aslı Süner Karakülah, Gamze Sarıkaya Uzan, Cagatay Gunay, Didem Soydemir, özlem Özsoy*
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1.099eP Neurofilament study: assessment of neurofilament levels in premature newborns

Marie Machiels[1], *Tamara Dangouloff, Nadège Hennuy, Vincent Rigo, Laurent Servais*
[1]*University of Liège, Liège, Belgium*

1.083P From CSF proteomics to functional models: validation of SMA biomarkers in C. elegans

Pamela Santonicola, Chiara Panicucci, Giada Onorato, Maria Bolla, Chiara Fiorillo, Delia Gagliardi, Stefania Corti, Angela Chambery, Elia Di Schiavi, ***Claudio Bruno***[1,2]
[1]*Centre of Translational and Experimental Myology, IRCCS Istituto Giannina Gaslini, Genova, Italy,* [2]*Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health-DINOGMI, University of Genova, Genova, Italy*

1.084P Cell-free plasma proteomics reveal multi-dimensional biomarker signatures in spinal muscular atrophy: insights from the RHU SMART cohort

Thomas Chetot, Arnaud Jacquier, Mazelin Laetitia, Zelia Chauvaud, Laurent Servais, Shams Ribault, Laurent Schaeffer, ***Carole Vuillerot***[1,2]
[1]*Hospices Civils De Lyon, Lyon, France,* [2]*PGNM, Université Claude Bernard Lyon 1, Lyon, France*

1.085P Efficient cellular and nuclear uptake and BBB crossing drive durable multi-organ rescue by DG9-PMO in SMA mice

Rika Maruyama[1], *Umme Sabrina Haque, Melissa Kohut, Jillian Claerhout, Razoan Al Rimon, Jun Ren, Md Nur Ahad Shah, Hong M. Moulton, Zamaneh Kassiri, Simon Gosgnach, Toshifumi Yokota*
[1]*University Of Alberta, Edmonton, Alberta, Canada*

1.086P Integration of SMN2 structures and variants in the clinical platform of CuidAME in Spain: analysis of 276 SMA patients

Andres Eduardo Nascimento Osorio[1], *Laura Blasco-Pérez, Stephanie Lotz Esquivel, Mar Costa-Roger, Jordi Leno, Maria Jesica Expósito Escudero, Rocio Garcia-Uzquiano, Laura Carrera Garcia, Daniel Natera de Benito, David Gomez Andres, Juan Francisco Vazquez-Costa, Laura Gonzalez Mera, Maria del Mar Garcia Romero, Rocio Calvo Medina, Miguel Lafuente, Raquel Blanco, Yolanda Martin, Cristina Puig-Ram, Eduardo Tizzano, . CUIDAME investigators Group*
[1]*Sant Joan de Déu Hospital, Barcelona, Spain*

1.087P Using long-read sequencing to decipher the SMN2 locus in severely affected SMA patients

Maureen Jacob[1,2,3], *Martin Pavlov, Elisabeth Graf, Sandy Lösecke, Michal Fischer, Omar Keritam, Cetin Hakan, Martin Krenn, Heike Kölbel, Ulrike Schara-Schmidt, Simone Thiele, Maggie C Walter, Andreas Ziegler, Juliane Winkelmann, Astrid Blaschek, Sebastian Eck, Katharina Vill, Matias Wagner*
[1]*Institute of Human Genetics, School of Medicine and Health, Technical University of Munich, Munich, Germany,* [2]*Institute of Neurogenomics, Helmholtz Munich, Neuherberg, Germany,* [3]*Department of Pediatric Neurology and Developmental Medicine, Ludwig Maximilian University of Munich (LMU), Hauner Children's Hospital , Munich, Germany*

Poster Session 1: 1.100eP- 1.104eP, 1.088P-1.090P: SMA – Treatments

1.100eP Clinical pharmacology of apitegromab in spinal muscular atrophy

Giridhar Tirucherai, Yang Xu, Nathalie Gosselin, Samer Mouksassi, Kathleen Koeck, Richard Riese, Chris Simard, Guochen Song, Doreen Barrett, Mouhamed Gueye, ***Jing Marantz***[1]
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1.101eP Phase 1 interim results evaluating salanersen safety, tolerability, pharmacokinetics, and exploratory efficacy for spinal muscular atrophy (SMA)

Stephanie Fradette[1], *Andreas Ziegler, Thomas Crawford, Valeria Sansone, Katarzyna Kotulska-Jóźwiak, Diana Castro, Peng Sun, Sara Alexanian, Jennifer Inra, Carrie Rubel, Julie Czerkowicz, Alissa Verone-Boyle, Wenjing Li, Shirley Wu, Yingying Liu, Richard Foster, Julia Heo, Leeann Burton*
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1.102eP Final results from the JEWELFISH study: 5-year risdiplam treatment in non-treatment-naïve individuals with spinal muscular atrophy (SMA)

Claudia Chiriboga[1], *Claudio Bruno, Tina Duong, Dirk Fischer, Janbernd Kirschner, Mariacristina Scoto, Eugenio M Mercuri, Valerie Schlegel, Ksenija Gorni, Heidemarie Kletzl, Pooneh Pordeli, Eleni Gaki, Francis Warren, Francesco Muntoni*
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1.103eP The SOLAR study: phase 3 study to evaluate the efficacy and safety of salanersen in participants aged 15–60 years with spinal muscular atrophy (SMA)

Stephanie Fradette[1], *Perry Shieh, Tim Hagenacker, Anna Kostera-Pruszczyk, Colleen O'Connell, Orla Hardiman, Lauren Elman, Peng Sun, Sara Alexanian, Jennifer Inra, Julie Czerkowicz, Natalie Schmitz, Shirley Wu, Alissa Verone-Boyle, Giulia Gambino, Yingying Liu, Richard Foster, Leeann Burton, Julia Heo*
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1.104eP STELLAR phase 3 studies to evaluate the efficacy and safety of salanersen in infants with spinal muscular atrophy (SMA)

Stephanie Fradette, Thomas Crawford, ***Crystal Proud***[1], *Richard Finkel, Eugenio Mercuri, Laurent Servais, Michelle Farrar, Peng Sun, Julia Heo, Sara Alexanian, Jennifer Inra, Carrie Rubel, Julie Czerkowicz, Alissa Verone-Boyle, Shirley Wu, Natalie Schmitz, Yingying Liu, Richard Foster, Ben Tichler, Leeann Burton*
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1.088P Real-world effectiveness of risdiplam in spinal muscular atrophy: a single-center experience compared with pivotal trials

Anna Lemska[1], *Sylwia Studzinska, Maria Mazurkiewicz-Beldzinska*
[1]*Medical University of Gdansk, Gdansk, Poland*

1.089P Intravenous and intrathecal onasemnogene abeparvovec for presymptomatic and symptomatic spinal muscular atrophy (SMA): long-term follow-up

Basil Darras[1], *Michelle A. Farrar, Eugenio M. Mercuri, Kevin A. Strauss, John W. Day, Yin-Hsiu Chien, Riccardo Masson, Roberto Bernardo Escudero, Iulian Alecu, Nigel Dodd, Lesa Mehl, Anne M. Connolly*
[1]*Boston Children's Hospital, Harvard Medical School, Boston, United States*

1.090P Final results from RESPOND: study of nusinersen in children with spinal muscular atrophy previously treated with onasemnogene abeparvovec

Crystal Proud[1], *Richard Finkel, John Brandsema, Riccardo Masson, Nancy Kuntz, Julie Parsons, Richard Foster, Wenjing Li, Ross Littauer, Judith Anesi, Ankur Sharma, Nicole Harris, Stephanie Fradette, Bora Youn*
[1]*Children's Hospital of The King's Daughters, Norfolk, VA, United States*

Short Oral Presentations 1: SP01 – SP05, Congenital myopathies, mechanisms and therapies

SP01 Loss of adenylosuccinate synthase (ADSS) isoenzymes impairs neuromuscular development in zebrafish

Ankita Suhas Tulangekar[1], *Elodie Costin, Anna Quach, Dean Campelj, Cara Timpani, Avnika Ruparelia, Emma Rybalka*
[1]*Victoria University, Melbourne, Australia*

SP02 Mutation-independent MyoAAV knockdown-and-replace gene therapy rescues muscle function in ACTA1 nemaline myopathy mice: a therapeutic platform for neuromuscular disease

Daniel Bouch[1,2], *Arun Paripati, Jingting Zhu, Yuchen Wu, Afrooz Rashnonejad*
[1]*Nationwide Children's Hospital, Columbus, Ohio, United States,* [2]*The Ohio State University, Columbus, Ohio, United States*

SP03 The replacement kinetics of the giant muscle protein nebulin are slow

Sylvia Bogaards[1], *Michaela Yuen, Yeszamin Onderwater, Carlos Clara, Ricardo Galli, Miguel Vizoso, Stefan Conijn, Eva Peters, Leila Nahidiazar, Nimo Annor, Julia Muldoon, Neil Wodd, Michael Previs, Kees Jalink, Jacco van Rheenen, Henk Granzier, Coen Ottenheijm*
[1]*Amsterdam UMC, Department of Physiology, Amsterdam, Netherlands*

SP04 Developing a patient-centric humanised young mouse model of congenital myopathy using iPSC-derived skeletal muscle progenitor cells

Darcy Pirotta[1,2], *Joshua Clayton, Omar Elaskalani, Connor McPhan, Gina Ravenscroft, Raelene Endersby, Rhonda Taylor*
[1]*Harry Perkins Institute of Medical Research, Perth, Australia,* [2]*University of Western Australia, Perth, Australia*

SP05 Testing allele-selective antisense oligonucleotide therapeutics in RYR1-related myopathy patient-derived muscle cells

Rhonda Taylor[1,2], *Jordan Crane, Jeremy Garcia, Connor McPhan, Anna Johnstone, Nigel Laing, Joshua Clayton, Gina Ravenscroft*
[1]*Harry Perkins Institute of Medical Research, Western Australia, Perth, Australia,* [2]*University of Western Australia, Western Australia, Perth, Australia*

Short Oral Presentations 2: SP06 – SP09, Toxic and inflammatory myopathies

SP06 Cellular mechanisms of mtDNA management and inflammation in mitochondrial myopathies

Emily Cross[1], *Marton Szabo, Daniel Lagos, Rita Horvath*
[1]*University Of Cambridge, Cambridge, United Kingdom*

SP07 Glucagon-like peptide-1 receptor agonist-associated myopathy: a metabolic double hit

Bryce Chang[1], *Nicolas Madigan, Margherita Milone, Marcus Pinto, Ashley Santilli, Teerin Liewluck*
[1]*Mayo Clinic, Rochester, United States*

SP08 Chemokine-associated inflammatory signatures in inclusion body myositis

Yusuke Baba[1,2], *Shinichiro Hayashi, Francia Victoria De Los Reyes, Shunsuke Funaguma, Aritoshi Iida, Satoru Noguchi, Steven A. Greenberg, Ichizo Nishino*
[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry (NCNP), Kodaira, Tokyo, Japan,* [2]*Department of Neurology, Graduate School of Medicine, The University of Tokyo, Bunkyo, Tokyo, Japan*

SP09 Immune checkpoint inhibitor–related myotoxicity: clinical spectrum, predictors of severity, and development of a pragmatic prognostic score

Sami Tayb-Boulahfa[1], *Lucas Pacoureau, Marie-Claire Bretagne, Sarah Léonard-Louis, Beatrice Labell, Eric Ballot, Marie-Alexandra Alyanakian, Rana Al Kouri, Olivier Benveniste, Joe-Elie Salem, Yves Allenbach*
[1]*Centre de Recherche en Myologie, UMRS 974, INSERM-Sorbonne University, Paris, France*

Short Oral Presentations 3: SP10 – SP15, DMD treatments

SP10 Mutation-independent upregulation of utrophin through CRISPR-based strategies for Duchenne muscular dystrophy

Mario Amendola[1], *Paola Galbiati, Maëlle Ralu, Simon Guiraud, D Luclerc, S Amistadi, Sumitava Dastidar, A Miccio, Francesco Saverio Tedesco*
[1]*Genethon, Evry, France*

SP11 Systemic delivery of full-length dystrophin using myotropic/split-intein AAV vectors leads to superior functional outcomes compared with micro-dystrophin

Jeffrey Chamberlain[1], *Theodore Reyes, Erynn Johnson, Timothy McMillen, Michael Regnier, James Ervasti, Hichem Tasfaout*
[1]*University of Washington School of Medicine, Seattle, United States*

SP12 RIPPLE ultrasound-mediated delivery platform enables non-invasive, immune-stealth delivery of full-length dystrophin ameliorating DMD phenotype in rodent disease model

Ivan Krivega[1], Charles Levine, Katelyne Benthall, Margarita Krivega, Brooke Babineau, Glenn Pacheco, Elizabeth del Greko, David Kim, Anagha Rao, Dale George, Nana Ingram, Yoko Takashima, Erin Tom, Gwendolyn Austin, Daniel Barajas, Tanaya Ghosh, Ramzi J Khairallah, Nancy Jorgesen, Steven Feinstein, Kenneth Greenberg
[1]SonoThera, South San Francisco, United States

SP13 Impact of C4BPA on muscle progenitor cell differentiation: insights for Duchenne muscular dystrophy treatment

Esther Fernandez-Simon[1], Ainoa Tejedera-Villafranca, Xiomara Fernandez-Garabay, James Clark, Panagiotis Katsikis, Priyanka Mehra, Andrew Galloway, Alexandra Monceau, Elisa Villalobos, Dan Cox, Javier Ramon-Azcon, Juan M Fernandez-Costa, Jordi Diaz-Manera
[a]John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom,

SP14 Givinostat reduces decline of contractile cross-sectional area and decreases fat infiltration in Duchenne muscular dystrophy

Krista Vandenborne, Rebecca Willcocks, Paolo Bettica, Sara Cazzaniga, Federica Alessi, Eugenio Mercuri[1]
[1]Pediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, Rome, Italy

SP15 Sustained functional, biomarker, and MRI therapeutic effects following GNT0004 gene therapy in ambulant boys with Duchenne muscular dystrophy: long-term phase 1/2 results from the GNT-016-MDYF trial

Francesco Muntoni, Vincent Laugel, Jean-Baptiste Davion, Silvana De Lucia, Manon Sanz, Laurine Buscara, Stephanie Blaie, Marie Sagot, Alexandra Barbier, Estelle Creoff, Nathalie Daniele, Eric Guemas, Minh-Dan Barthelemy, Amanda Riviere, Florence Masson, Anne Gobert, Serge Braun, Arnaud Valent, Fei Cao[1], Gerald Perret
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Poster Session 2: 2.001eP-2.002eP, 2.001P-2.003P: Becker muscular dystrophy

2.001eP Rationale, study design and baseline characteristics in the GRAND CANYON clinical trial of sevasemten in Becker muscular dystrophy adults

Craig McDonald[1], *Richard Roxburgh, Eugenio Mercuri, Kristl Claeys, Michela Guglieri, Erik H. Niks, Katherine Mathews, Anne M. Connolly, Luca Bello, Barry Byrne, Aravindhan Veerapandiyan, Jim MacDougall, Roxana Donisa Dreghici, Joanne Donovan*
[1]*University of California, United States*

2.002eP Patient and public involvement in Becker muscular dystrophy: establishment and outcomes of a nationwide patient group in Japan

Hiroyuki Shibasaki[1,2], *Masaru Torigoe, Hikaru Endo, Yoshitsugu Aoki, Akinori Nakamura, Hirofumi Komaki, Tamotsu Takeda*
[1]*Japan Muscular Dystrophy Association, Tokyo, Japan,* [2]*National Center of Neurology and Psychiatry, Tokyo, Japan*

2.001P Urine titin levels in patients with Becker muscular dystrophy differ substantially according to DMD deletion pattern

Hiroyuki Awano[1], *Akinori Nakamura, Kayo Osawa, Taku Shirakawa, Yoshinori Nambu, Ryosuke Bo, Suyoshi Matsumura, Michinori Funato, Akiko Wakisaka, Satoshi Kuru, Yasuhiro Takeshima, Tatsuharu Sato, Michio Kobayashi, Kazuma Sugie, Masafumi Matsuo*
[1]*Organization for Research Initiative and Promotion, Tottori University, Yonago, Japan*

2.002P In-frame DMD exon deletions in male participants from the all of us cohort without a diagnosis of dystrophinopathy

Daniele Sabbatini, Riccardo Iannaco, Luca Vedovelli, Dario Gregori, Elena Pegoraro, ***Luca Bello***[1]
[1]*Department of Neurosciences, Dns, University of Padova, Padova, Italy, Padova, Italy*

2.003P Long-term stabilization of function in Becker: sevasemten prevented functional decline up to 3.5 years in MESA open-label extension

Craig McDonald[1], *Erik H. Niks, Han Phan, Hani Kushlaf, Diana Castro, Katherine Mathews, Arun S. Varadhachary, Anne M. Connolly, Michela Guglieri, Doris Leung, Jeffrey Statland, Rosaline Quinlivan, Varun Sreenivasan, Aravindhan Veerapandiyan, Nicholas E. Johnson, Brenda Wong, James Signorovitch, Jess Marden, Mirko Fillbrunn, Joanne Donovan*
[1]*University of California, United States*

Poster Session 2: 2.003eP-2.009eP, 2.004P-2.012P: CMD - Clinical care and case reports

2.003eP Thromboembolic burden and stroke risk in pediatric LMNA-related congenital muscular dystrophy: implications for preventive anticoagulation

Andres Eduardo Nascimento Osorio[1], *Sergi Cesar, Laura Carrera, Agustin Lujan, Carlos Ortez, Jesica Exposito, Josefina Garcia, Silvia Cerezo, Stephanie Lotz, Irene zschaeck, Andrea Greco, Nuria Diez, Patricia Cerralbo, María Vila, Chiara Gatnau, Berta Estevez, Jordi Pijuan, Ruben Berroecos, Daniel Natera De Benito, Georgia Sarquella Brugada*
[1]*Hosp Sant Joan De Deu, Neuromuscular unit, Neurology department , Barcelona, Spain*

2.004eP Expanding clinical and genetic spectrum of alpha-dystroglycanopathies: single-center experience

Didem Ardıçlı[1], *Ezgi Erem, Busranur Cavdarli, Aysegul Nese Citak Kurt, Cavidan Nur Semerci Gunduz*
[1]*Ankara Bilkent City Hospital, Ankara, Turkey*

2.005eP Study of motor and respiratory function in Ullrich congenital muscular dystrophy in Japan

Kumiko Ishiguro[1], *Yuki Kihara, Yuko Motohashi, Hirofumi Komaki, Takatoshi Sato, Minobu Shichiji, Keiko Ishigaki*
[1]*Tokyo Women's Medical University, Tokyo, Japan*

2.006eP A novel homozygous INPP5K mutation in siblings with congenital cataract and muscular dystrophy

Serkan Kirik[1], *Zehra Nur Ağ, Hazel Mahmat, Yavuz Ataş, Ömer Bektaş*
[1]*Elazig Fethi Sekin City Hospital Division of Pediatric Neurology, Elazig, Turkey*

2.007eP Baseline data from pediatric patients with LAMA2 related dystrophies enrolled in a prospective and longitudinal natural history study in France

Marjorie Banos, Erwan Gasnier, Abdennour Chellah, Melina Brindeau, Aurelie Phelep, Dominique Duchene, Frederic Fer, Marta Gomez Garcia de la Banda, Pierre Dupont, Soraya Houmel, Carole Vuillerot, Shotaro Tachibana, Sabrina Oumnia, Ulrike Walther-Louvier, Souad Touati, Lea Thevenet, Saadane Kirouani, Susana Quijano Roy, ***Andreea Seferian***[1]
[1]*Institute of Myology, I-Motion clinical trials platform, Assistance Publique des Hôpitaux de Paris (AP-HP), Hôpital Armand Trousseau, Paris, France*

2.008eP Compound heterozygous POMT1 mutations in a boy with muscular dystrophy-dystroglycanopathy C1

Dilek Cavusoglu[1], *Basak Gokcay, Omer Lutfi Koyuncu, Gizem Nur Izgi*
[1]*Departments of Pediatric Neurology, Afyonkarahisar Health Sciences University, Afyon, Turkey*

2.009eP Clinical presentation of a patient with TRIP4-associated congenital muscular dystrophy
Tetiana Shkliarska[1], *Maryna Patsora, Nataliia Olkhovych, Nataliia Samonenko, Svitlana Kormoz, Natalia Gorovenko*
[1]*National Children's Specialized Hospital "Ohmatdyt" Of The Ministry Of Health Of Ukraine, Kyiv, Ukraine*

2.004P Phenotypic expansion of COL12A1 related myopathy: insights from a multigenerational family
My Vuong Hermansen[1,2,3], *Cecilie Fremstad Rustad, Kristian Tveten, Lena Lande Wekre, Kristin Ørstavik*
[1]*Norwegian Centre for Rare Diseases, Nesodden, Norway,* [2]*Section for Rare Neurological disorders and EMAN, Department of Neurology, Oslo University Hospital, Oslo, Norway,* [3]*Institute of Clinical Medicine, Faculty of Medicine, University of Oslo, Oslo, Norway*

2.005P Clinical and genetic spectrum of congenital muscular dystrophy in Turkey: a large multicenter cohort
Mustafa Kömür[1], *Ezgi Caglar, Didem Soydemir, Uluc Yis, Neslihan Ozcan, Ozlem Herguner, Didem Ardıclı, Aysegül Nese Cıtak Kurt, Haluk Topaloglu, Ayse Nur Coskun, Deniz Yuksel, Pınar Saka, Gokcen Oztuncer, Gamze Sarıkaya Uzan, Bihter Sen Sahin, Esra Sarıgecilli, Hulya Maraslı, Sevim Turay, Yavuz Atas, Study Group*
[1]*Mersin University Faculty of Medicine, Mersin, Turkey*

2.006P Clinical and genetic profile of children with LAMA2-related dystrophies in Turkey
Didem Ardıçlı[1], *Busranur Cavdarli, Hatice Bektas, Uluc Yis, Olcay Unver, Ozlem Herguner, Mustafa Komur, Gamze Sarikaya Uzan, Gulcin Akinci, Gokcen Oz Tuncer, Nezir Ozgun, Serkan Kirik, Hulya Maras Genc, Ilknur Erol, Serdar Pekuz, Didem Soydemir, Deniz Yuksel, Beril Talim, Haluk Topaloglu, Turkish LAMA2 Study Group*
[1]*Ankara Bilkent City Hospital, Ankara, Turkey*

2.007P Preventing early mortality in COL6-RD: motor trajectories and proactive respiratory monitoring in a national cohort (n=190)
Diana Abramova[1], *Svetlana Artemyeva, Sergey Bardakov, Ludmila Kuzenkova, Anastasia Monakhova, Eugenia Melnik, Sergey Nikitin, Irina Shulyakova, Dmitry Vlodavets, Sergey Kurbatov*
[1]*ANO "Ullrich- Bethlem Myopathy", Nizhny Novgorod, Russia*

2.008P Noncontrolled, nonblinded, multicenter, collaborative phase 2 trial of steroid therapy for Fukuyama congenital muscular dystrophy
Takatoshi Sato[1], *Terumi Murakami, Harumasa Nakamura, Hisateru Tachimori, Takami Ishizuka, Hideki Oi, Mari S Oba, Kumiko Ishiguro, Minobu Shichiji, Yuki Kihara, Yasuhiro Takeshima, Mariko Taniguchi Ikeda, Ayako Hattori, Yuko Shimizu Motohashi, Hiroyuki Awano, Ryosuke Bo, Keiko Ishigaki*
[1]*Department of Pediatrics, Tokyo Women's Medical University, Tokyo, Japan*

2.009P Clinical and genetic spectrum of Emery–Dreifuss muscular dystrophy (EDMD) from India
Nimita Rani[1], *Farsana Mustafa, Alisha Reyaz Reyaz, William L Macken, Ashwin Dalal, Lindsay A Wilson, ICGNMD Consortium, Rohit Bhatia, Ajay Garg, Robert D S Pitceathly, Kumarasamy Thangaraj K, M V Padma Srivastava, Michael Hanna, Venugopalan Y Vishnu*
[1]*Department of Neurology, All India Institute of Medical Sciences, New Delhi, India, New Delhi, India,*

2.010P Establishing clinical trial readiness in patients age 0-5 years with LAMA2-related congenital muscular dystrophy (READY CMD LAMA2)
Lindsay Alfano[1], *Kevin Warf, Megan Iammarino, Natalie Reash, Linda Lowes, Mina Jain, Sara Marshall, Rejuan Haque, Kaitlin Batley, Hoda Abdel Hamid, Katherine Mathews, Perry Shieh, Carolina Tesi-Rocha, Cuixia Tian, Craig Zaidman, Carsten Bonnemann, Guy Brock, Megan Waldrop, A. Reghan Foley, Anne Connolly*
[1]*Nationwide Children's Hospital, Columbus, United States*

2.011P Prevalence of anxiety and depression in young adults with chronic neuromuscular disease: a call for scheduled screening
Hoda Abdel-Hamid[1], *Ahmed Hamza, Adam Yassin*
[1]*University of Pittsburgh, Pittsburgh, United States*

2.012P Genetic variants associated with neuromuscular disorders in a large cohort of heart transplant recipients due to cardiomyopathy
Anna Benterud[1], *Trine Haug Popperud, Fredrik Hellem Stenseth, Kjell-Arne Arntzen, Kristina Ingrid Helena Hermann Haugaa, Kristin Ørstavik*
[1]*Section for Rare Neuromuscular Disorders and EMAN, Department of Neurology, Oslo University Hospital, Institute of Clinical Medicine, Faculty of Medicine, University of Oslo, Oslo, Norway*

Poster Session 2: 2.013P-2.016P: CMD – Physiopathology

2.013P MEF2A modulation as a therapeutic strategy for LAMA2-related muscular dystrophy
Julie Yue Yuan[1], *Veronica Pini, Stefano Carlo Previtali, Francesco Muntoni*
[1]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom*

2.014P In vitro characterization of the extracellular matrix and basement membrane in LAMA2-related congenital muscular dystrophies

Louise Benarroch[1], *Lauren Struble, Kamel Mamchaoui, Anne Bigot, Nur Villar-Quiles, Tanya Strojkovic, Valérie Allamand*
[1]*Centre de Recherche en Myologie - Umrs974, Paris, France*

2.015P Advanced human iPSC-based modelling of LMNA-related congenital muscular dystrophy enables development of targeted genetic therapies for muscle laminopathies

Daniel Moore, Heather Steele-Stallard, Luca Pinton, Valentina Lionello, Lucia Rossi, Artadokht Aghaeipour, Salma Jalal, Cherry Tsz Yan Wong, Angela Clara-Hwang, Gisele Bonne, Peter Zammit, ***Francesco Saverio Tedesco***[1,2,3]
[1]*Department of Cell and Developmental Biology, University College London, London, United Kingdom,* [2]*The Francis Crick Institute, London, United Kingdom,* [3]*Dubowitz Neuromuscular Centre, Great Ormond Street Hospital and UCL Great Ormond Street Institute of Child Health, London, United Kingdom*

2.016P Improving the diagnosis of collagen VI-related dystrophies (COL6-RDs) with collablots

Nadia Osegui Barcenilla[1], *Solange Kapetanovic García, Usua Laresgoiti, Andrea López Martínez, Virginia Arechavala Gomeza*
[1]*Nucleic Acid Therapeutics for Rare Diseases (NAT-RD), Biobizkaia Health Research Institute, Barakaldo, Spain*

Poster Session 2: 2.010eP-2.028eP, 2.017P-2.039P: DMD - clinical care and case reports

2.010eP Barriers to delivery of standard-of-care physiotherapy for Duchenne muscular dystrophy: a nationwide cross-sectional survey of physiotherapists and caregivers

Gulcin Akinci[1], *Eray Ontas, Numan Bulut, Ipek Alemdaroglu-Gurbuz, Oznur Tunca, Haluk Topaloglu*
[1]*UHS Izmir Faculty of Medicine, Dr. Behcet Uz Children's Hospital, Pediatric Neurology, Izmir, Turkey, Izmir, Turkey*

2.012eP Development of an Italian registry to assess natural history in Duchenne muscular dystrophy: a next generation EU–PNRR study

Marika Pane[1], *Daniela Leone, Claudia Brogna, Beatrice Berti, Concetta Palermo, Luca Kunsztler, Sonia Messina, Valeria Ada Maria Sansone, Antonio Di Muzio, Sophia Paolucci, Romina Venditti, Giorgia Coratti, Eugenio Maria Mercuri*
[1]*Catholic University, Centro Clinico Nemo Pediatrico Policlinico A.Gemelli , Rome, Italy*

2.013eP Tolerability of givinostat in combination with exon-skipping therapy in individuals with Duchenne muscular dystrophy

Cameron Wade[1,2], *Andrew Bramer, Paul Horn, Alexander Zygmunt, Lisa Reebals, Meghan McAnally, Cuixia Tian*
[1]*Cincinnati Children's Hospital Medical Center, Cincinnati, United States,* [2]*University of Cincinnati College of Medicine, Cincinnati, United States*

2.014eP Machine learning prediction of non-invasive ventilation initiation in Duchenne muscular dystrophy

Rageen Rajendram[1,2], *Abishek Bhuvanaratnam, Tithi Paul, Laura McAdam, Reshma Amin*
[1]*Holland Bloorview Kids Rehabilitation Hospital, Toronto, Canada,* [2]*University of Toronto, Toronto, Canada*

2.015eP Hydrotherapy in Duchenne muscular dystrophy, a systematic review

Christian De Goede[1,2], *Candiss Argent, Eva Thackeray, Kelly Bowden-Davies, Christopher Morse*
[1]*Lancashire Teaching Hospitals NHS Trust, Preston, United Kingdom,* [2]*Manchester Metropolitan University, Manchester, United Kingdom*

2.016eP Leveraging human-centered and systems design to advance early product development for lower limb contracture management in Duchenne muscular dystrophy

Theofilos Kempapidis[1], *Oisin Minogue, Hayley Philippault, David Buckley, Emily Reuben, Alexandra Johnson, Michael Bond, Farnaz Nickpour*
[1]*University Of Liverpool, Liverpool, United Kingdom*

2.017eP Fatal fat embolism syndrome without recognized fracture in patients with Duchenne muscular dystrophy: two case reports

Cameron Wade[1,2], *Chet Villa, Larry B Goldstein, Cuixia Tian, Alexander Zygmunt, Kathryn Rice, Laura Bagwell, Eimear McGovern, David N Toupin*
[1]*Cincinnati Children's Hospital Medical Center, Cincinnati, KY, United States,* [2]*University of Cincinnati College of Medicine, Cincinnati, KY, United States*

2.019eP Impact of functional mobility on dental aesthetic index (DAI) scores in children with Duchenne muscular dystrophy - a single centre experience

Vineeth Jaison[1], *Roshan Reji, Hemiya George, Joe Mathew*
[1]*Christian Medical College and Hospital, Ludhiana, India*

2.020eP Long-term follow-up of communication methods utilizing residual functions in patients with advanced-stage Duchenne muscular dystrophy
Kotaro Arioka[1], *Eri Yawatari, Masatoshi Ishizaki*
[1]*Kumamoto Saishun Medical Center, Kumamoto, Japan*

2.021eP School attendance in Duchenne muscular dystrophy
Gokcen Oz Tuncer[1], *Gülbahar Kurt Bayır, Ömer Gökhan Çelik, Ayşe Aksoy*
[1]*Department of Pediatric Neurology, Ondokuz Mayıs University Faculty of Medicine ,Samsun, Turkey*

2.022eP Genetic and clinical spectra of dystrophinopathies in Turkish children
Muhammet Gultekin Kutluk, ***Gokcen Oz Tuncer***[1], *Hülya Maraş Genç, Aysel Ünal, Ayşegül Yılmaz, Güven Toksoy, Duygu Güner Özcanyüz, Ayşe Aksoy, Naciye Füsun Toroman*
[1]*Ondokuz Mayıs Üniversitesi Pediatric Neurology, samsun, Turkey*

2.023eP Evaluation of the psychosocial status and quality of life of healthy siblings of children diagnosed with Duchenne muscular dystrophy
Ömer Demir, ***Aysima Özçelik***[1], *Baran Çalışgan*
[1]*Gaziantep University, Gaziantep, Turkey*

2.024eP Pan-immune-inflammation value predicts earlier loss of ambulation in Duchenne muscular dystrophy
Bilge Özgör[1], *Işınsu Bıçakcıoğlu, Mahir Tan, Gül Yücel, Meral Karadağ, Serdal Güngör*
[1]*İnönü University, Malatya, Turkey*

2.025eP Clinical, pathological, and genetic characteristics of x-linked dilated cardiomyopathy caused by DMD mutation
Meirong Liu[1], *Na Ji, Xiaoke Wu, Wanyu Xu, Qi Fang*
[1]*The First Affiliated Hospital Of Soochow University, Suzhou, China*

2.026eP Establishing the diagnosis of dystrophinopathy in a female manifesting carrier: a multimodal diagnostic approach
Monique Driesssen van Haeringen[1], *Anders Oldfors, Kalliopi Sofou, Carola Hedberg-Oldfors*
[1]*Department of Pediatric Neurology, Queen Silvia Children's Hospital, Sahlgrenska University Hospital, Gothenburg, Region Västra Götaland, Sweden*

2.027eP Post delandistrogene moxeparvovec-rokl immune mediated myositis in Duchenne muscular dystrophy patient with DMD 10-43 deletion
Maria Moredock, Michelle Lee, Meganne Leach, Kylee Kastelic, Brittany Gurgel, ***Erika Finanger***[1]
[1]*Oregon Health and Science University, Portland, United States*

2.028eP Advanced cardiac therapies in the dystrophinopathy spectrum: a review of two-cases
Zurisadai Gonzalez Castillo[1], *Laura Radel, Erica Knoll, Leslie Nelson, Kaitlin Batley*
[1]*UTSW Medical Center/Children's Health, Dallas, United States*

2.017P Patient preferences, barriers and facilitators regarding physical activity in Duchenne muscular dystrophy
Evelien Fleerakkers[1,2], *Isabelle Pirson, Anna Stuurman, Hermien E Kan, Erik H Niks, Menno van der Holst*
[1]*Leiden University Medical Center, Leiden, Netherlands,* [2]*Duchenne Center Netherlands, Leiden, Netherlands*

2.018P Upper limb function and spinal surgery outcome in non-ambulatory patients with Duchenne muscular dystrophy- a single paediatric centre experience
Nicola Burnett[1], *Lianne Abbott, Victoria Crook, Sophie Burton, Jose Longatto, Shira Rabinowicz, Adnan Manzur, Francesco Muntoni, Anna Sarkozy*
[1]*Great Ormond Street Hospital, London, United Kingdom*

2.019P Lived experiences of lower-limb stretching in Duchenne muscular dystrophy: family challenges and practical adaptations
Theofilos Kempapidis[1], *Oisin Minogue, Hayley Philippault, David Buckley, Emily Reuben, Alexandra Johnson, Michael Bond, Farnaz Nickpour*
[1]*University Of Liverpool, Liverpool, United Kingdom*

2.020P Premature school disruption in Duchenne: a marker of societal participation in India
Himshweta Balwani[1,2,3], *Sneha Shyamprasad, Gauri Khandekar, Sharanya Ravindra, Ann Mathew*
[1]*Bangalore Baptist Hospital, Bangalore, India,* [2]*Synapse Neuro and Child Development Centre, Bangalore, India,* [3]*Virgo Health care, Bangalore, India*

2.021P Neuropsychiatric comorbidities in Duchenne muscular dystrophy: association with brain dystrophin isoforms

Dragana Vucinic[1], *Ana Kosac, Jelena Mladenovic, Minja Ninkovic, Marko Jerinic, Milos Brkusanin, Jovan Pesovic, Dusanka Savic Pavicevic, Vedrana Milic Rasic*

[1]*Clinic of Neurology and Psychiatry for Children and Youth, Belgrade, Serbia*

2.022P Duchenne muscular dystrophy in Indonesia: real-world data from a tertiary neuromuscular center

Kristy Iskandar[1], *Ery Kus Dwianingsih, Farah Shahnaz Pravinovia, Dhea Ananda, Gunadi*

[1]*Division of Neurology Department of Child Health Faculty of Medicine, Public Health and Nursing/ UGM Academic Hospital, Universitas Gadjah Mada, Yogyakarta, Indonesia*

2.024P Early diagnosis of dystrophinopathies and other primary muscle disorders in patients presenting with hypertransaminasemia/hyperCKemia

Carlos Ignacio Ortez Gonzalez[1,2], *Jesica Exposito, Estephie Lotz, Silvia Cerezo, Laura Carrera, Natera de Benito Daniel, Berta Estévez Arias, Andrés Nascimento*

[1]*Neuromuscular Unit, Sant Joan de Déu Hospital, Barcelona, Spain,* [2]*Applied Research in Neuromuscular Diseases, Institut de Recerca Sant Joan de Déu, Barcelona, Spain*

2.025P Clinical and molecular features of Duchenne muscular dystrophy in Senegal: insights from a West African cohort

Mame Diarra Dia[1], *Henriette V.F. Senghor, Rokhaya Diagne, Alassane Diop, Aloïse Benoît Diouf, Marie Emilie Yandé Ndong, Marietou Traoré, Maimouna Santos, Khalifa Ababacar Mbaye, Lala Bouna Seck, Mamadou sy, Maouly Fall, Adjaratou Dieynaba Sow, Ngor Side Diagne, Sergi Beltrán Agullo, Andres Nascimiento, Daniel Natera, Loreto Martorell, Moustapha Ndiaye, Pedro M Rodriguez Cruz*

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2.026P Identifying when to initiate use of the performance of upper limb in ambulant Duchenne muscular dystrophy

Olivia Martineau[1], *Lianne Abbott, Georgia Stimpson, Giovanni Baranello, Nicola Burnett, Rachel Thomas, Jose Longatto, Amy Wolfe, Anna Sarkozy, Adnan Manzur, Francesco Muntoni*

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2.027P Complex duplications in the DMD gene: prevalence and detection using combined long-read and transcriptomic sequencing

Stefan Nicolau[1,2], *Daniel Koboldt, Alayne Meyer, Breanna Trecha, Eleni Drakou, Emma Frair, Leigh Gabel, Diane Dunn, Robert Weiss, Kevin Flanigan*

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2.028P Feasibility and preliminary findings of the Duchenne muscular dystrophy -educational accessibility and participation tool: an Indian pilot study

Gauri Khandekar[1], *Sneha Banavara Shyamprasad, Sharanya P Ravindra, V Jaison, Himshweta Balwani, Ann Agnes Mathew*

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2.029P Improving cardiac care for Duchenne muscular dystrophy in the UK through a collaborative approach

Catherine Turner[1], *Sadie Bennett, Caroline Coats, Adam McDiarmid, Alex Johnson, Lisa Kuhwald, Kadhim Kadhim, Adam Kerr, Lynne Williams, Michela Guglieri, John Bourke*

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2.030P Advancing lower limb stretching in Duchenne muscular dystrophy; how do evidence and clinical practice align?

Theofilos Kempapidis[1], *Oisin Minogue, Hayley Philippault, David Buckley, Emily Reuben, Alexandra Johnson, Michael Bond, Farnaz Nickpour*

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2.031P Aerobic and strength activity in daily life in patients with Duchenne muscular dystrophy

Evelien Fleerakkers[1,2], *Anna Stuurman, Hermien Kan, Erik Niks, Menno van der Holst*

[1]*Leiden University Medical Center, Leiden, Netherlands,* [2]*Duchenne Center Netherlands, Leiden, Netherlands*

2.032P What it means to live with Duchenne muscular dystrophy in sub-Saharan Africa: a descriptive study from Senegal

Mame Diarra Dia[1], *Henriette VF Senghor, Rokhaya Diagne, Alassa Diop, Alouis Benoit Diouf, Seynabou Dieng, Marietou Traore, Khalifa Ababacar Mbaye, Lala Bouna Seck, Mamadou Sy, Maouly Fall, Adjaratou Sow, Ngor Side Diagne, Andres Nacimiento, Daniel Natera, Loreto Martorell, Andoni Urtizberea, Frande Leturcq, Moustapha Ndiaye, Pedro M Rodriguez Cruz*

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2.033P Physical activity and the relation with mental well-being in Duchenne muscular dystrophy

*Evelien Fleerakkers, Anna Stuurman, Hermien Kan, Rosanne Govaarts, Erik Niks, **Menno Van Der Holst**[1,2]*
[1]Leiden University Medical Center, Leiden, Netherlands, [2]Duchenne Center Netherlands, Leiden, Netherlands

2.034P Predicting the 95th centile stride velocity (SV95C) at 6-, 12-, 18-, and 24-months in steroid-treated boys with Duchenne muscular dystrophy

***Georgia Stimpson**[1], Guillaume Parinello, Céline Cluzeau, Margaux Poleur, Niculina Butoianu, Jana Haberlova, Sherry Kodsy, Madalina Leanca, Damjan Osredkar, Anna Potulska-Chromik, Lena Szabo, Francesco Muntoni, Silvana De Lucia, Rachida-Lydie Zanfongnon, Arnaud Valent, Fei Cao, Gerald Perret, Damien Eggenspieler, Laurent Servais, on behalf of the ActiLiege Next Study Group*
[1]MDUK Oxford Neuromuscular Centre and NIHR Oxford Biomedical Research Centre, University of Oxford, Oxford, United Kingdom

2.035P Hydrotherapy for health in boys and adolescents with Duchenne muscular dystrophy, an inpatient control study

***Christian De Goede**[1,2], Candiss Argent, Kelly Bowden Davies, Gladys Onambele-Pearson, Christopher Morse*
[1]Lancashire Teaching Hospitals NHS Trust, Preston, United Kingdom, [2]Manchester Metropolitan University, Manchester, United Kingdom

2.036P "Walk like an Egyptian" Comparing the natural history of Egyptian and European Duchenne muscular dystrophy patients in a prospective multicentre study

***Sherry Kodsy**[1], Areej Saleh, Georgia Stimpson, Guillaume Parinello, Céline Cluzeau, Margaux Poleur, Niculina Butoianu, Jana Haberlova, Madalina Leanca, Damjan Osredkar, Anna Potulska-Chromik, Lena Szabo, Damien Eggenspieler, Nicolas Bovy, Laurent Servais*
[1]Galaa Military Medical Complex, Cairo, Egypt

2.037P Integrated protein and transcript analysis reveals mechanisms affecting dystrophin levels in in-frame DMD deletions

***Emma Frair**[1], Robert Weiss, Gabrielle Dufresne, Xiaojin Zhang, Leigh Gabel, Sarah Atkins, Diane Dunn, Alayne Meyer, Sang Cheol Seok, Russell Butterfield, Stefan Nicolau, Kevin Flanigan*
[1]Center for Gene Therapy, Abigail Wexner Research Institute, Nationwide Children's Hospital, Columbus, United States

2.038P Population-based newborn screening scoping study for Duchenne muscular dystrophy in New South Wales and the Australian capital territory, Australia

***Kristi Jones**[1], Sarah Shin, Kirsten Boggs, Jessica Lu, Merly Babu, Julie Curtin, Lyndal Douglas, Michelle Farrar, Rosie Junek, Didu Kariyawasam, Edwin Kirk, Eric Law, Michelle Lorentzos, David Mowat, Shelley Pirreca, Enzo Ranieri, Jacqui Russell, Tiffany Wotton, Gladys Ho, Bruce Bennetts*
[1]Sydney Children's Hospitals Network - Westmead, Sydney, Australia

2.039P Predicting exon skipping therapy outcomes using natural history of exon 44 and 45 skipping-equivalent genotypes

***Eleni Drakou**[1], Alayne Meyer, Leigh Gabel, Ezgi Saylam, Megan Waldrop, Stefan Nicolau, Kevin Flanigan*
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Poster Session 2: 2.029eP-2.031eP, 2.040P-2.052P: DMD - imaging and outcome measures

2.029eP Defining plantar pressure distribution in children with Duchenne muscular dystrophy and its associations with ambulation

***Numan Bulut**[1], Ali Yalçin, Semra Topuz, İpek Alemdaroğlu-Gürbüz, Göknur Haliloğlu, Öznur Tunca*
[1]Hacettepe University, Faculty of Physical Therapy and Rehabilitation, Department of Physiotherapy and Rehabilitation, Ankara, Turkey

2.030eP Development and validation of a novel dystrophinopathy foot index (DFI) for comparison and surgical decision-making related foot contracture

***Meghan Moore Burk**[1], Amanda Appel, Denesh Ratnasingam, Jeri Forster, Jason Rhodes, Anne Stratton, Carolyn Kelley, Susan Apkon*
[1]Children's Hospital Colorado, Aurora, United States

2.031eP Evaluation of the efficacy of viltolarsen to Duchenne muscular dystrophy using muscular imaging

***Satoshi Kuru**[1], Makoto Minamiyama, Takahiro Nakayama, Toshio Saito, Tsuyoshi Matsumura*
[1]Suzuka National Hospital, Suzuka, Japan

2.040P A pathophysiological inflection point in Duchenne muscular dystrophy uncovered by myofibre density analysis: implications for therapeutic strategies

***Tetsuhiro Yamakado**[1,2], Yuka Ishikawa, Yusuke Ishida, Koki Ise, Lei Wang, Yoshitaka Oda, Masumi Tsuda, Taichi Kimura, Masayoshi Nagao, Manabu Ito, Shinya Tanaka, Zen-ichi Tanei*
[1]Department of Pathology, National Hospital Organization Hokkaido Medical Center, Sapporo, Japan, [2]Department of Cancer Pathology, Faculty of Medicine, Hokkaido University, Sapporo, Japan

2.041P Development and content validation of a novel educational accessibility and participation tool for Duchenne muscular dystrophy in India

Sneha Banavara Shyamprasad[1], *Himshweta Balwani, Vineeth Jaison, Gauri Khandekar, Sharanya P Ravindra, Ann A Mathew*
[1]*Synapse Neuro Center, Bengaluru, India*

2.042P Using centiles for the north star ambulatory assessment in Duchenne muscular dystrophy: a prospective real-world study

Amy Wolfe[1,2], *Georgia Stimpson, Anna Mayhew, Adnan Manzur, Deborah Rideout, Anna Sarkozy, Jasmine Watts Whent, Lianne Abbott, Sarah Gregson, Lindsey Pallant, Jennie Sheehan, Alex Schofield, Hayley Ramjattan, Francesca Henderson, Evelin Milev, Francesco Muntoni, Giovanni Baranello, Study Group NorthStar Clinical Network*
[1]*Great Ormond Street Hospital, London, United Kingdom,* [2]*University College London, London, United Kingdom*

2.043P Summarising lower limb contractures affecting gait and function in ambulant boys with Duchenne muscular dystrophy

Kate Carroll[1,2], *Katy de Valle, Justine Adams, Peng Zhi, Ian Woodcock, Eppie Yiu, Rachel Kennedy*
[1]*The Royal Children's Hospital, Melbourne, Melbourne, Australia,* [2]*Murdoch Children's Research Institute, Melbourne, Australia*

2.044P Evidence of longitudinal validity of Duchenne video assessment 2.0, a home-based clinical outcome assessment for Duchenne muscular dystrophy

Marielle Contesse[1], *Gary Gensler, Mary Hubbard, Sure Radhiv Kumar, Richard Barnes, Dorys Marian, Mindy Leffler*
[1]*Red Nucleus, Yardley, United States*

2.045P Longitudinal kinematic gait changes in Duchenne muscular dystrophy: a pilot study on disease progression and therapy effects

Pablo Ruiz, Guy Cheron, Ana-Maria Cebolla, Valentine Tahon, ***Nicolas Deconinck***[1,2,3]
[1]*NeuroMove, Laboratory of Movement Neurophysiology, Université Libre de Bruxelles, Brussels, Belgium,* [2]*Hôpital Universitaire des Enfants Reine Fabiola (HUDERF, HUB), Brussels, Belgium,* [3]*Neuromuscular Reference Center (NMRC) UZ Gent, Ghent, Belgium*

2.046P Novel measures of functional movements in patients with Duchenne muscular dystrophy and spinal muscular atrophy: correlation with cardiopulmonary outcomes

Rebecca Scharf[1], *Casey Litle, Arafat Rahman, Ariful Islam, Chelsea Masterson, Leila Orszag, Silvia Blemker, Laura Barnes*
[1]*University Of Virginia, Charlottesville, United States*

2.047P Using a novel index of neural control to mechanistically understand cognition in carrier women of dystrophin mutations

Varsha Penumalee[1], *Hane Chen, Md Moinul Ahsan, Aaron James Kaat, Le Kang, Mathula Thangarajh*
[1]*Virginia Commonwealth University, Richmond, VA, United States*

2.048P Hospitalizations at pediatric institutions among Duchenne and Becker muscular dystrophy: ICU use, mortality, and cost utilizing the PHIS database

Emily Hayes[1], *Deipanjan Nandi, Linda Cripe, Grace Paul, Richard Shell, Courtney Gelpi, Kavitha Kotha, L. Kate Lamberta, Ezgi Saylam, Lydia Wright*
[1]*Nationwide Children's Hospital, Columbus, United States*

2.049P Further validation of the DuMAND checklist in Duchenne muscular dystrophy: normative data, longitudinal analysis, and cross-cultural validation

Eline Cuveele[1,2], *Sam Geuens, Jente Dreezen, Kim Wittebols, Joanna Willen, Céline R. Gillebert, Liesbeth De Waele*
[1]*University Hospitals Leuven, Leuven, Belgium,* [2]*KU Leuven, Leuven, Belgium*

2.050P Quantitative MRI, valuable surrogate biomarkers in monitoring disease progression of DMD, an analysis of Genethon's multicentric natural history study with standardized follow-up

Fei Cao[1], *Vincent Laugel, Silvana De Lucia, Jean-Baptiste Davion, Laure Le Goff, Michaela Guglieri, Caroline Espil, Brigitte Chabrol, Tristan Montier, Andres Nascimento, Nuria Muelas, Nicolas Deconinck, Liesbeth De Waele, Benjamin Marty, Harmen Reyngoudt, Rachida-Lydie Zanfongnon, Eric Guemas, Gerald Perret, Arnaud Valent, Francesco Muntoni*
[1]*Genethon, Evry, France*

2.051P A prognostic score for time to loss of hand-to-mouth ability in Duchenne muscular dystrophy (DMD)

Utkarsh Dang, Nicolae Done, Shuang Wang, James Signorovitch, Jessica R. Marden, Heather Gordish-Dressman, Taylor Wang, Fangzhou Wei, Susan J. Ward, ***Craig McDonald***[1]
[1]*University Of California Davis, Sacramento, United States*

2.052P Connectivity biosignatures of cognitive challenges in Duchenne muscular dystrophy

Mathula Thangarajh[1], *James Bjork, Liangsuo Ma*
[1]*Virginia Commonwealth University, Richmond, United States*

2.033eP Pre-implementation safety validation of governance-controlled AI for communication of emerging therapies in Duchenne muscular dystrophy: a givinostat proof-of-concept

Kirandeep Sahajpal[1]
[1]*Good Hope Hospital, Birmingham, United Kingdom*

2.034eP Two-year motor function outcomes following treatment with delandistrogene moxeparvovec-rokl in young boys with Duchenne muscular dystrophy: A single center experience

Kaitlin Batley[1,2], *Leslie Nelson, Erica Knoll, Danielle Forrest, Madeline Brown, Melanie Valle, Zurisadai Gonzalez Castillo*
[1]*UT Southwestern Medical Center, Dallas, United States,* [2]*Children's Health, Dallas, United States*

2.035eP Therapeutic novelties in Duchenne muscular dystrophy – experience from a European expert center for rare neurological diseases

Mihaela Cioclu[1], *Cristina Suteu, Tania Corina Mboh, Mihaela Amelia Dobrescu, Mihaela Adela Vintan, Diana Gabriela Barca, Ioana Minciu, Carmen Burloiu, Cristina Haritina Motoescu, Carmen Sandu, Niculina Butoianu*
[1]*Alexandru Obregia Clinical Psychiatry Hospital, Bucharest, Romania*

2.036eP Assessing side effects of thrombocytopenia and hypertriglyceridemia for individuals with Duchenne muscular dystrophy treated with givinostat

Paul Horn[1,2], *Lisa Reebals, Alexander Zygmunt, Andy Bramer, Meghan McAnally, Cameron Wade, Cuixia Tian*
[1]*Cincinnati Children's Hospital Medical Center, Cincinnati, United States,* [2]*College of Medicine, University of Cincinnati, Cincinnati, United States*

2.037eP Safety of viltolarsen in patients with Duchenne muscular dystrophy in real-world clinical practice interim analysis of post-marketing surveillance in Japan

Akari Kawano[1], *Tomoyuki Nakagawa, Yoshinori Hayase, Sari Horiuchi, Tatsuya Suzuki, Isao Kashimori, Kazutaka Takagaki*
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2.038eP Real-world evidence of viltolarsen in Japanese boys with Duchenne muscular dystrophy: four-year results from the remudy-DMD registry

Harumasa Nakamura[1], *Mari Oba, Shoki Izumi, Hakami Ishizuka, Hirofumi Komaki, Remudy-DMD Investigator Group*
[1]*National Center Of Neurology And Psychiatry, Tokyo, Japan*

2.039eP Real world four-year outcomes on the efficacy and safety of viltolarsen for Duchenne muscular dystrophy

Eri Takeshita[1], *Yuko Shimizu-Motohashi, Takatoshi Hara, Hirofumi Komaki*
[1]*National Center Of Neurology And Psychiatry, Tokyo, Japan*

2.040eP QTc distribution in Duchenne muscular dystrophy patients: implications for therapies with potential cardiac effects

Eugenio Mercuri[1], *Maddalena Bianchetti, Camilla Silvij, Sophia Paolucci, Federica Mirra, Romina Venditti, Elena Sofia Spanò, Paolo Galletti, Angelica Bibiana Delogu, Marika Pane*
[1]*Pediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, Rome, Italy*

2.041eP Trajectory of performance of upper limb in patients with Duchenne muscular dystrophy treated with givinostat relative to a modeled natural history cohort

John Brandsema[1], *Jessica R. Marden, Jonathan Freimark, Katherine A. O'Connor, Andrea Parodi, Elyse Swallow, Emma Billmyer, Michel Michaëls, Menno van der Holst, Saskia Houwen, James Signorovitch, Paolo Bettica, Erik H. Niks*
[1]*Children's Hospital of Philadelphia, Perelman School of Medicine at the University of Pennsylvania, Philadelphia, Pennsylvania, United States*

2.042eP Hematological effects of vamorolone in boys with Duchenne muscular dystrophy (DMD) – comparison with prednisone

Nicolas Deconinck[1], *Lenka Jurikova, Eric Hoffman, Raoul Rooman, Elisabeth Genestin, Giovanni Baranello*
[1]*Paediatric Neurology HUB - Hôpital Universitaire des Enfants Reine Fabiola ULB, Brussels, Belgium*

2.043eP Comprehensive panel of pharmacokinetic, pharmacodynamic, and biomarker measures drives clinical development of BMN 351 for Duchenne muscular dystrophy

Lexa Staskus[1], *Kevin Larimore, Pavan Chityala, Kristin Obrochta-Moss, Huiyu Zhou, Katja Hebestreit, Rob Power, Evan Witt, Laura Wetzel, Maria Abbasi, Annie Greenslade, Ara Yu, Kristen Jahr, Peter Velazquez, Andrew Melton*
[1]*Biomarin Pharmaceutical Inc., San Rafael, CA, United States*

2.044eP Pooled safety analysis from phase 1 to phase 3 clinical trials of delandistrogene moxeparvovec in Duchenne muscular dystrophy

Crystal Proud[1], *Craig M. McDonald, Eugenio M. Mercuri, Francesco Muntoni, Craig M. Zaidma, Vannary Pizzo, Matthew Furgerson, Jim Jin, Alexander P. Murphy, Mark Vivien, Marianne Gerber, Jerry R. Mendell*

[1]*Children's Hospital of the King's Daughters, Norfolk, United States*

2.045eP FORZETTO, a phase 3 study of z-rostudirsen in ambulatory males with DMD mutations amenable to exon 51 skipping

Iva Holder[1], *Doug Kerr, Xiaoyan Yin, Katy Meilleur, Jordan Messer, Gabriel Helmlinger, Prashant Bansal, Soma Ray, Monica Fay, Maria Naylor*

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2.047eP Pharmacokinetics and pharmacodynamic effects of the POLARIS-101 (TM) rationally designed, next-generation capsid

Matthew Harmelink[1], *Jamie Marshall, Glen Banks, Jessie Hanrahan, Paul Herzich, Nicolas Christoforou, Patrick Gonzalez, Gabriel Brooks*

[1]*Solid Biosciences, Charlestown, United States*

2.048eP Development of a therapeutic approach for Duchenne muscular dystrophy based on the glycosyltransferase Fut8

Nozomi Hayashiji[1], *Tomohiko Fukuda, Jianguo Gu, Yoshitsugu Aoki, Eri Arikawa-Hirasawa*

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2.049eP Enhancing antisense oligonucleotide efficacy in Duchenne muscular dystrophy by histone deacetylase inhibitors drug repurposing

Ramla Omar[1], *Rachele Rossi, Francesco Muntoni*

[1]*Institute of Child Health, UCL, London, United Kingdom*

2.050eP Efficacy and safety of early intervention with delandistrogene moxeparvovec in DMDMDX mice

Nicki Pukos, John Snedeker, Betsy Pray, Ellyn Nesbit, ***Rachael Potter***[1]

[1]*Sarepta Therapeutics, Inc., Cambridge, United States*

2.051eP Xanthine oxidoreductase inhibition improves cellular energetics and mitochondrial function in metabolically and/or structurally stressed muscle

Satomi Shirakaki[1], *Yuki Ashida, Norio Motohashi, Naoyuki Kamatani, Yoshitsugu Aoki*

[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

2.052eP CRISPR-Cas9-mediated upregulation of utrophin ameliorates Duchenne muscular dystrophy

Paola Galbiati, D Luclerc, S Amistadi, A Miccio, ***Mario Amendola***[1,2,3]

[1]*Genethon, Evry, France,* [2]*inserm, Evry, France,* [3]*University of Foggia, Foggia, Italy*

2.053eP Real-world characteristics and care patterns of DMD patients in Japan and the United States: a medical claims data comparison on the same platform

Tatsuya Suzuki[1], *Tsubasa Bito, Masahito Oishi, Kakeru Nakagawa, Akari Kawano, Takuya Toramoto, Hidenori Komori, Ryotaro Sonoda, Takeshi Tanaka*

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2.054eP Safety and efficacy of viltolarsen in DMD therapy. Data from 4 years of real clinical practice in Russia

Natalia Balamut[1], *Dmitry Vlodavets, Svetlana Artemyeva, Olga Glebovskaya, Fatima Nakhusheva, Dina Ayzatulina, Egor Grankin, Svetlana Ipatova, Elena Shishkina, Dar'ya Korotkova, Olga Zhivaeva, Elena Sayfullina, Galina Treskina, Dmitry Gukosyan, Ger Momot, Larisa Tsyngunova*

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2.055eP Validation lab: allowing standardized in vitro and in vivo experiments for candidate treatments for Duchenne muscular dystrophy

Tiberiu Loredan Stan[1], *Davy Van De Vijver, Christa Tanganyika-de Winter, Anne Poos, Annemieke Aartsma-Rus*

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2.056eP Positive preliminary liver safety profile using next-generation capsid POLARIS-101 in the SGT-003 gene therapy program for Duchenne muscular dystrophy

Matthew Harmelink[1], *Jamie Marshall, Nicolas Christoforou, Patrick Gonzalez, Gabriel Brooks*

[1]*Solid Biosciences, Charlestown, United States*

2.057eP Duchenne hub UK network enabling effective and efficient clinical trial set up and delivery

Emma Heslop[1], *Mike Bond, Alexandra Johnson, Emily Reuben, Phillip Cammish, Jessica Elliott, Megan McNiff, Giovanni Baranello, Jon Hastie, Anne-Marie Childs, Volker Straub, Michela Guglieri*
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2.058eP Safety and efficacy of viltolarsen treatment in patients with Duchenne muscular dystrophy: a retrospective study with 5-year follow-up

Michinori Funato[1], *Reina Iwata, Ryoya Uno, Misaki Yumioka, Yoshiko Kato, Yuji Yamada*
[1]*NHO Nagara Medical Center, Gifu, Japan*

2.059eP Muscle composition and muscle injury biomarkers improve with BMN 351 treatment

Laura Wetzel, Kristin Obrochta Moss, ***Kevin Larimore***[1], *Katja Hebestreit, Dan Gaffney, Hai Liu, Maria Abbasi, Lexa Staskus, Ishnoor Sidhu, Janine Ilagan, Evan Witt, Ling Liu, Chris Rayner, Rob Power, Monika A. Sigg*
[1]*BioMarin, San Rafael, United States*

2.060eP Generation of iPSC lines with a pathogenic DMD pseudoexon mutation c.9225-285A>G

Emma Niba[1], *Hiroshi Koide, Masafumi Matsuo*
[1]*Laboratory of Molecular and Biochemical Research, Juntendo University, Tokyo, Japan*

2.061eP Real-world patient and treatment characteristics in children under 4 years old with Duchenne muscular dystrophy receiving viltolarsen

Robert Crozier[1], *Mara Wood, Denise B. Quigley, Max Jacobs, Michelle L. Previtera*
[1]*NS Pharma, Paramus, United States*

2.062eP Real-world patient and treatment characteristics in a non-ambulatory cohort ≥12 years old with Duchenne muscular dystrophy receiving viltolarsen

Denise Quigley[1], *Mara N Wood, Max Jacobs, Michelle L Previtera*
[1]*NS Pharma, Paramus, United States*

2.063eP Building muscle, bridging models: an integrated platform for Duchenne muscular dystrophy gene therapy

Reema Flynn-Rizk[1], *Margarita Egiian, Lisa Hoffman*
[1]*Western University, London, Canada*

2.064eP QTc prolongation in an individual with Duchenne muscular dystrophy treated with givinostat and lessons for clinicians

Cuixia Tian[1,2], *Alexander Zygmunt, Lisa Reebals, Cameron Wade, Paul Horn, Meghan McAnally, Benjamin Mantell, Chet Villa*
[1]*Cincinnati Children's Hospital Medical Center, Cincinnati, United States,* [2]*University of Cincinnati, College of Medicine, Cincinnati, United States*

2.065eP CRISPR activation of utrophin as a mutation-independent approach for Duchenne muscular dystrophy therapy

Maëlle Ralu, Simon Guiraud, Sumitava Dastidar, Paola Galbiati, Francesco Saverio Tedesco, ***Mario Amendola***[1,2,3]
[1]*Genethon, Evry, France,* [2]*inserm, Evry, France,* [3]*University of Foggia, Foggia, Italy*

2.067eP Efficacy and safety of golodirsen and casimersen compared with placebo in Duchenne muscular dystrophy (ESSENCE): phase 3 results and post hoc analysis

Francesco Muntoni[1,2], *Emma Ciafaloni, Fernando Chloca, Nicolas Deconinck, Ameneh Masud, Ihor Sehinovych, Kerri Drummond, Weijian Liu, James Signorovitch, Andrés Nascimento Osorio, Sheffali Gulati, Maria Judit Molnar, Maria Mazurkiewicz-Beldzinska, Eugenio Mercuri, Craig M. McDonald*
[1]*University College London, London, United Kingdom,* [2]*National Institute for Health Research Great Ormond Street Hospital Biomedical Research Centre, London, United Kingdom*

2.053P Interim pharmacokinetic and pharmacodynamic results of the phase 1/2a AVANCE1 trial on tricyclo-DNA-induced DMD exon 51 skipping in Duchenne muscular dystrophy

Helge Amthor[1,2], *Alessandra Pennisi, Tim Ulinski, Corinne Guitton, Simon Guiraud, Luis Garcia, A consortium*
[1]*Unité de Recherche PHARMAColigo UR20261, Université de Versailles Saint-Quentin-en-Yvelines, Versailles, France,* [2]*Hôpital Raymond Poincaré, Assistance Publique – Hôpitaux de Paris, Garches, France*

2.054P Investigating the role of EGFR signalling in muscle dystrophies: implications for Duchenne muscular dystrophy

Esther Fernandez-Simon[1], *Ainoa Tejedera-Villafranca, Xiomara Fernandez-Garabay, James Clark, Alexandra Monceau, Elisa Villalobos, Dan Cox, Javier Ramón-Azcón, Juan M Fernandez-Costa, Jordi Diaz-Manera*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom*

2.055P Long-term monitoring of cystatin C in boys with Duchenne muscular dystrophy undergoing viltolarsen therapy in real clinical practice

Tatiana Gremiakova[1], *Tatiana Morozova, Olga Gremiakova, Ekaterina Gorshkova, Olga Varnakhona, Andrey Stepanov*
[1]*Pediatric High-tech Neuromuscular Centre of Federal State Budget Institution «Central Clinical Hospital with Ambulance», Moscow, Russia*

2.056P SGT-003 gene therapy stabilizes the DAPC and improves muscle integrity in Duchenne muscular dystrophy

Patrick Gonzalez[1], *Stephen Bradley, Michael Lawlor, Tatyana Vetter, Brandon Chan, David Wolfson, Peter Nadeau, Kevin Whittlesey, Jamie Marshall, Jessie Hanrahan, Gabriel Brooks, Nicolas Christoforou*
[1]*Solid Biosciences, Charlestown, United States*

2.057P Real-world experience of safety and tolerability in 89 individuals with Duchenne muscular dystrophy initiating givinostat treatment; a retrospective review

Lisa Reebals[1], *Andrew Bramer, Paul Horn, Alexander Zygmunt, Meghan McAnally, Cuixia Tian*
[1]*Cincinnati Children's Hospital Medical Center, Cincinnati, United States*

2.058P Preliminary clinical cardiac findings using POLARIS-101, a next-generation capsid with high cardiac tropism, in the SGT-003 program

Patrick Gonzalez[1], *Jamie Marshall, Meghan Soustek-Kramer, Marie Ralff, Jessie Hanrahan, Nicolas Christoforou, Matthew Harmelink, Gabriel Brooks*
[1]*Solid Biosciences, Charlestown, United States*

2.059P Effect of vamorolone and prednisone on glucose and lipid metabolism in boys with Duchenne muscular dystrophy (DMD)

Lenka Jurikova[1], *Giovanni Baranello, Erik Hoffman, Raoul Rooman, Elisabeth Genestin, Nicolas Deconinck*
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2.060P Decoupling growth and loss of ambulation in Duchenne muscular dystrophy (DMD): Long-term outcomes with Vamorolone

Giovanni Baranello[1,2], *Erik H. Niks, Ekaterina Gresko, Aki Linden, Shabir Hasham, Eugenio Mercuri, Francesco Muntoni, Craig M. McDonald*
[1]*Dubowitz Neuromuscular Centre, UCL Gt Ormond St Inst Child Health, London, United Kingdom,* [2]*NIHR Gt Ormond St Hospital Biomedical Research Centre, London, United Kingdom*

2.061P Brogidirsen 5-year safety and efficacy outcomes in Duchenne muscular dystrophy patients: findings from an ongoing open-label extension study

Hirofumi Komaki[1], *Eri Takeshita, Katsuhiko Kunitake, Yuko Shimizu-Motohashi, Shinsuke Maruyama, Rena Mlyagi, Yan Li, Yoshitsugu Aoki*
[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

2.062P Delandistrogene moxeparvovec in Duchenne muscular dystrophy: functional and safety outcomes up to 3 years post-infusion in the EMBARK study

Jerry R. Mendell, Francesco Muntoni, Craig M. McDonald, Eugenio M. Mercuri, Emma Ciafaloni, ***Hirofumi Komaki***[1], *Carmen Leon-Astudillo, Andrés Nascimento, Crystal Proud, Ulrike Schara-Schmidt, Aravindhan Veerapandiyan, Craig M. Zaidman, Matthew Furgerson, Mark Vivien, Damon R. Asher, Alexander P. Murphy, Marianne Gerber, Carmen O. Torre, Marianna Manfrini, Louise R. Rodino-Klapac*
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2.063P Engineered muscle-targeting extracellular vesicles for systemic delivery of micro-dystrophin: novel redosable strategy for Duchenne muscular dystrophy

David Tran, Meena Murali, Christy Lau, Bailey Murphy, Huilan Lin, Firouz Mohsenian, Yujia Li, ***Mafalda Cacciottolo***[1], *Linda Marban, Kristi Elliott, Minghao Sun*
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2.064P Non-viral lipid nanoparticles enable plasmid micro-dystrophin delivery in Duchenne muscular dystrophy

Desiree Genovese[1], *Andrea Pirrottina, Luca Digiacomo, Marianna Cosentino, Ludovica Apa, Carmine Nicoletti, Antonio Musarò, Daniela Pozzi, Giulio Caracciolo*
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2.065P Sustained muscle function improvement after a single in vivo dose of antibody oligonucleotide conjugate (AOC) in the mdx mouse model for Duchenne muscular dystrophy

Tiberiu Loredan Stan[1], *Davy Van De Vijver, Christa Tanganyika-de Winter, Lizette van der Pijl, Anne Poos, Elias Post, Floris van Delft, Nazli Hilal Türkmen, Annemieke Aartsma-Rus*
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2.066P Phase 1/2 results of BMN 351, an antisense oligonucleotide for exon 51 skip-amenable Duchenne muscular dystrophy

Erik Niks[1], *Giovanni Baranello, Marcos Madruga-Garrido, Marika Pane, Valeria Sansone, Hai Liu, Peter Velazquez, Orli Rosen, Cigdem Ayanoglu, David Neil, Haluk Topaloglu*
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2.067P Primary results of the US expanded access protocol (EAP) study in vamorolone-treated DMD boys

Amy Harper[1], *Nancy Kuntz, Anna Roth, Ekaterina Gresko, Craig M. McDonald*
[1]*Department of Neurology, Children's Hospital of Richmond at VCU, Richmond, United States*

2.068P Dystrophic cardiac fibrosis is regulated by Msr1 via macrophage-associated TGF-ß1/SMAD signalling

Norio Motohashi[1], *Yoshitaka Mizobe, Hotake Takizawa, Maria Tsoumpra, Koichi Kimura, Yoshitsugu Aoki*
[1]*National Center of Neurology and Psychiatry (NCNP), Kodaira, Japan*

2.069P Discovery of a splicing variant of the myostatin gene and its application to the treatment of Duchenne muscular dystrophy

Masafumi Matsuo[1], *Atsuko Takeuchi, Kayo Osawa, Atsuhito Takeda, Kayoko Kanamitsu, Kazuhiro Maeta, Hisahide Nishio*
[1]*Kobe University, Kobe, Japan*

2.070P Specific peptides targeting the inhibitory core of myostatin alleviate pathologival phenotypes in dystrophin- and caveolin-3-deficient mice

Yutaka Ohsawa[1], *Masahiro Fujino, Shin-ichiro Nishimatsu, Yoshihide Sunada*
[1]*Kawasaki Medical School/Kurashiki, Kurashiki, Japan*

2.071P Analysis of the intracellular localization of exon-skipping ASO conjugates in human skeletal muscle cells

Laia Torres Masjoan[1,2], *David Coulson, Arta Aghaeipour, Jennifer Frommer, Sara Aguti, Haiyan Zhou, Francesco Muntoni*
[1]*Neurodegenerative Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom,* [2]*Genetics and Genomic Medicine Research and Teaching Department, UCL Great Ormond Street Institute of Child Health, London, United Kingdom*

2.072P Optimization of intramuscular administration and in-vivo monitoring of gene-editing therapy in Duchenne muscular dystrophy

Ozge Findik Sener[1,2], *Fanny Sage, Erik H. Niks, Marco C. De Ruiter, Marten A. Engelse, Nynke Verwoerd, Susi S. Rauh, Niels Geijsen, Mark Burgmans, Hermien E. Kan*
[1]*Leiden University Medical Center, Leiden, Netherlands,* [2]*The Novo Nordisk Foundation Center for Stem Cell Medicine (reNEW), Leiden, Netherlands*

2.073P Establishment of a multidisciplinary online network (BRIDGE-NMD / e-consult) for adverse event management in DMD gene therapy

Hirofumi Komaki[1], *Atsuhito Takeda, Mureo Kasahara, Ayano Inui, Toshihiro Sawai, Kohsuke Imai, Masafumi Onodera, Takayuki Kishi, Keiko Ishigaki*
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2.074P BASECAMP: a phase 2a dose comparison and exploratory efficacy study of SAT-3247 in ambulatory boys with Duchenne muscular dystrophy

Anne Connolly[1], *Ian Woodcock, Kristiana Salmon, Ryan Mitchell, Phil Lambert, Michael Rudnicki, Wildon Farwell*
[1]*Nationwide Children's Hospital, Columbus, United States*

2.075P TRAILHEAD: an open-label, phase 2 study evaluating long-term safety and efficacy of SAT-3247 in adults with Duchenne muscular dystrophy (DMD)

Anne Connolly[1], *Ian Woodcock, Gayatri Jain, Kristiana Salmon, Ryan Mitchell, Michael Rudnicki, Wildon Farwell*
[1]*Nationwide Children's Hospital, Columbus, United States*

2.076P MAGIC: a translational framework unifying human 3D models and gene therapy development for rare muscle diseases

Mariam Zouhair[1,2], *Sumitava Dastidar, Yunsong Jiang, Eugenia Carraro, Eduard Ayuso, Xabier Perez Garmendia, Sungwoo Choi, Giuseppe Ronzitti, Francesco Muntoni, Hildegard Büning, Mario Amendola, Andrea Serio, Francesco Saverio Tedesco, www.magic-horizon.eu*
[1]*The Francis Crick Institute, London, United Kingdom,* [2]*Department of Cell and Developmental Biology, University College London, London, United Kingdom*

2.077P Identifying muscle stem cell tropic AAV variants through directed evolution

Harry Chen[1], *Michael Emami, Courtney Young, Bradley Smith, Braeden Freitas, Thomas Rando, Oluwatayo Ikotun, April Pyle, Melissa Spencer*

[1]*University Of California, Los Angeles, Los Angeles, United States*

2.078P Pre-transplant rehabilitation improves donor cell engraftment efficiency in the skeletal muscle of Duchenne muscular dystrophy mouse model

Mayuho Miki[1], *Nana Takenaka-Ninagawa, Rukia Ikeda, Megumi Yokomizo-Goto, Clémence Kiho Yoshioka, Mio Iwasaki, Tomoki Aoyama, Hidetoshi Sakurai*

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2.079P Givinostat simplified weight-based dosing: rationale for minimized exposure differences and efficacy at different doses and body weights

Aravindhan Veerapandiyan[1], *Federica Alessi, Francesco Bellanti, Sara Cazzaniga, Paolo Bettica, Craig M McDonald, Eugenio Mercuri*

[1]*Department of Pediatrics, Division of Neurology, Arkansas Children's Hospital, Little Rock, United States*

2.080P Delandistrogene moxeparvovec micro-dystrophin expression and safety in 2–3-year-olds with Duchenne muscular dystrophy in ENDEAVOR and ENVOL studies

Craig M. McDonald, ***Francesco Muntoni***[1], *Andrea Gangfuss, Crystal Proud, Craig M. Zaidman, Laurent Servais, Anne M. Connolly, Patsy Thrasher, Mark Vivien, Alex Haile, Jennifer Thomas-Ahner, Jianfeng Meng, Alexander P. Murphy, Becca B. Zhang, Ewa Karasek, Pooneh Pordeli, Louise R. Rodino-Klapac*

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2.081P Open-label extension analysis suggests givinostat delays age at loss of ambulation in patients with Duchenne muscular dystrophy

David Gómez Andrés, Valeria Sansone, Andrea Parodi, Odile Boespflug-Tanguy, Natalie Goedeker, Paolo Bettica, Tracey Willis, ***Eugenio Mercuri***[1]

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2.082P cGAS-STING as a novel therapeutic target in Duchenne muscular dystrophy

Russell Rogers[1], *Alice Rannou, Nancy Manriquez, Sankalpa Chakraborty, Adarsh Balaji, Jonah Zarrow*

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2.083P Oral N-acetylglucosamine improves muscle function and provides additional benefit over prednisolone alone in a murine model of Duchenne muscular dystrophy.

Sachiko Sato[1], *Guillaume St-Pierre, Ann Rancourt, Maude Fillion, Masahiko S. Satoh*

[1]*Faculty of Medicine, Laval University, Quebec, Canada*

2.084P Interim safety results from an open-label extension study of givinostat treatment for Duchenne muscular dystrophy

John Brandsema[1], *Gyula Acasdi, Federica Alessi, Paolo Bettica, Odile Boespflug-Tanguy, Michela Guglieri, Erika L. Finanger*

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2.085P Update on the INSPIRE DUCHENNE phase 1/2 study of the next-generation microdystrophin gene therapy candidate SGT-003 for Duchenne muscular dystrophy

Eugenio Mercuri[1], *Aravindhan Veerapandiyan, Kevin Flanigan, Craig McDonald, Crystal Proud, Perry Shieh, Hernan Gonorazky, Alicia Henriquez, Craig Zaidman, Nancy Kuntz, Renata Shih, Erika Finanger, Patrick Gonzalez, Matthew Harmelink, Gabriel Brooks*

[1]*Pediatric Neurology Catholic University and Nemo Pediatrico Fondazione Policlinico Gemelli IRCCS, Rome, Italy*

2.086P Neuronal targeted AAV micro-dystrophin restores neurobehaviour and brain dystrophin interactors in mdx52 mouse

Konstantina Tetorou[1,2], *Monica Rebeca Gil-Garzon, Darren Chambers, Wing Sum Chu, Simon Waddington, Alex Kavanagh, Nicha Songsilph, Joanne Ng, Francesco Muntoni*

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[2]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, and NIHR Biomedical Research Centre, London WC1N 3JHL, London, United Kingdom,*

2.087P Mechanisms of mineralocorticoid receptor antagonist efficacy in the dystrophic skeletal muscle microenvironment

Jill Fortney[1], *Swathy Krishna, Reva Reddy, Christoph Lepper, Peyton Debell, Jeovanna Lowe, Zhen-Ru Alice Liu*

[1]*The Ohio State University College of Medicine, Columbus, United States*

2.088P Zeleciment rostudirsen increased dystrophin protein levels and demonstrated functional improvement in clinical measures in the DELIVER trial
Ian Woodcock, Susan Apkon, Nicolas Deconinck, Liesbeth De Waele, Kevin Flanigan, Hugh McMillan, Marika Pane, Stefano Previtali, ***Perry Shieh***[1]*, Prashant Bansal, Soma Ray, Soma Ray, Dazhe Wang, Douglas Kerr, Maria Naylor, Michela Guglieri*
[1]*University of California, Los Angeles, United States*

2.129P Development of Next-Generation AAV.U7snRNA-Based Gene Therapy Vectors for Exon 17 Skipping to Treat Duchenne Muscular DystrophyPap
Liubov Gushchina[1,2]*, Taylor Ferguson, Nehaal Patrick, Gabrielle Dufresne, Kayla Stevens, Hui Lin, Kevin Flanigan*
[1]*Jerry R. Mendell Center for Gene Therapy, Nationwide Children's Hospital, Columbus, United States,* [2]*Department of Pediatrics, The Ohio State University Wexner Medical Center, Columbus, United States*

Poster Session 2: 2.068eP-2.072eP, 2.089P-2.105P: Dystrophinopathies

2.068eP Preclinical to human translation of regenerative index: quantification of progressive muscle degeneration and disease-modifying treatment response
Ryan Mitchell[1]*, Johnathan Smid, Charis McPherson, Shanti Rayagiri, Jacob Monast, Steven Moore, Frank Gleeson, Phil Lambert, Wildon Farwell, Michael Rudnicki*
[1]*Satellos Bioscience Inc., Toronto, Canada*

2.069eP Dystrophin-deficient skeletal muscle is further stiffened by contraction-induced injury but is resilient to isometric fatigue
Deirdre Merry[1,2]*, Pavithran Devananthan, Rebecca Craven, Kellie Joe, Gretel Major, Jiayi Chen, Natalia Kabaliuk, Angus Lindsay*
[1]*University Of Otago, Christchurch, New Zealand,* [2]*University of Canterbury, Christchurch, New Zealand*

2.070eP Real-time kinetic monitoring of sarcolemmal fragility in patient-derived DMD muscle-on-a-chip
Ainoa Tejedera-Villafranca[1]*, Martín Ruiz-Gutiérrez, Petra Bauzon, Maria J Ugarte-Orozco, Armando Cortés-Reséndiz, Mehdi Torabi, Javier Ramón-Azcón, Juan M Fernández-Costa*
[1]*Institute for Bioengineering of Catalonia (IBEC), Barcelona, Spain*

2.071eP Epilepsy in dystrophinopathies: prevalence, electroclinical features, and brain dystrophin isoforms involvement in a large paediatric cohort
Francisco Agustin Cerra[1]*, Adolfo Gallo, María Eugenia Foncuberta, Bernabé López Caparrós, Roberto Caraballo, Soledad Monges*
[1]*Hospital de Pediatría "Prof. Dr. Juan P. Garrahan", Ciudad Autónoma De Buenos Aires, Argentina*

2.072eP Early cardiac dysfunction in mdx/Utrn−/− mice as a severe preclinical model for Duchenne muscular dystrophy
Tomonori Kitaura[1]*, Kentaro Hashikami, Ryotaro Hori, Yasunori Nio*
[1]*Axcelead Drug Discovery Partners, Inc., Fujisawa, Japan*

2.089P Brain-specific dystrophin isoforms have a function in radial glia during human cerebral cortex organoid development
Chaitra Sathyaprakash[1]*, Katsuhiko Kunitake, Tatsuo Mano, China Tatebori, Reiko Terada, Tadafumi Hashimoto, Daisuke Kawauchi, Toshiyuki Araki, Momoko Watanabe, Mariko Taniguchi-Ikeda, Hideya Sakaguchi, Yoshitsugu Aoki*
[1]*Department of Molecular Therapy, National Center for Neurology and Psychiatry, Kodaira, Tokyo, Japan*

2.090P Anesthetic considerations for repeated imaging in DMD rat models
Margarita Egiian[1,2]*, Reema Flynn-Rizk, Susan Tyler, Karman Lehal, Chantal Villeneuve, Lisa Hoffman*
[1]*University Of Western Ontario, London, Canada,* [2]*Lawson Research Institute, London, Canada*

2.091P 3D FN-silk scaffolds supports generation of human induced pluripotent stem cell derived cardiomyocyte models for Becker muscular dystrophy
Petter Nhi[1]*, Carolina Åstrand, My Hedhammar, Cristina Al-Khalili Szigyarto*
[1]*KTH Royal Institute of Technology, Stockholm, Sweden*

2.092P Decoding contraction-dependent pathology in in vitro human three-dimensional DMD tissues
Carolina Rodríguez-Gallo[1]*, Timothée Caboche, Chiara Ninfali, Juan Manuel Fernandez-Costa, Javier Ramon Azcon*
[1]*IBEC, Barcelona, Spain*

2.093P Establishment of a microminipig model of Duchenne muscular dystrophy with human disease–like phenotypes and its application to gene therapy
Itsumi Sato[1,2]*, Michihiro Imamura, Hichem Tasfaout, Atsuhito Takeda, Takashi Okada, Jeffrey Chamberlain, Yoshitsugu Aoki*
[1]*Department of Molecular Therapy, National Institute of Neuroscience, National Center of Neurology and Psychiatry (NCNP), Tokyo, Japan,* [2]*Department of Pediatrics, Faculty of Medicine and Graduate School of Medicine, Hokkaido University, Sapporo, Japan*

2.094P Tregs suppress fibrosis in muscular dystrophy

*Philip Farahat, Gerald Coulis, Diego Jaime, **Sergio Villalta**[1]*
[1]University Of California Irvine, Irvine, United States

2.095P Endocrine-associated decoupling of pancreatic function drives glucose intolerance in the mdx mouse model of Duchenne muscular dystrophy

Angus Lindsay[1,2,3,4]*, Gretel Major, Joel Eliades, Jiayi Chen, Charles Najt*
[1]University of Otago, Christchurch, New Zealand, [2]University of Canterbury, Christchurch, New Zealand, [3]Biomolecular Interaction Centre, Christchurch, New Zealand, [4]Maurice Wilkins Centre for Molecular Biodiscovery, Auckland, New Zealand

2.096P Multi-omics characterisation of skeletal muscle-derived vesicles reveals Duchenne muscular dystrophy-associated signatures

Francesco Catapano[1,2,3]*, Darren Chambers, Abas Laftah, Micol Falabella, Rahul Phadke, Prashant Srivastava, Costanza Emanueli, Francesco Muntoni*
[1]The Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom, [2]National Institute for Health Research, Great Ormond Street Institute of Child Health Biomedical Research Centre, University College London, London, United Kingdom, [3]Dubowitz Neuromuscular Centre, Division of Neuropathology, UCL Queen Square Institute of Neurology, London, United Kingdom

2.097P Functional impact of DMD Fibro-adipogenic progenitors in engineered 3D human skeletal muscle tissues

*Chiara Ninfali, Xiomara Fernández-Garibay, Ainoa Tejedera-Villafranca, Elia López-Serrano, Esther Fernández-Simón, Jordi Díaz-Manera, Javier Ramón-Azcón, **Juanma Fernández-Costa**[1]*
[1]Institute for Bioengineering of Catalonia (IBEC), The Barcelona Institute of Science and Technology (BIST), Barcelona, Spain

2.098P Gut microbiota dysbiosis drives altered immune responses and muscle pathology in Duchenne muscular dystrophy

Debora Mostosi[1]*, Andrea Farini, Federica Perillo, Aitor Blanco-Míguez, Antonella De Palma, Veronica Marrella, Paola Maragno, Davide Perico, Mirella Meregalli, Marzia Belicchi, Davide Bazzani, Monica Molinaro, Francesco Strati, Federica Facciotti, PierLuigi Mauri, Barbara Cassani, Nicola Segata, Yvan Torrente*
[1]University of Milan, Milan, Italy

2.099P 3D tissue engineered skeletal muscle to model dystrophinopathies and determine the efficacy of exon skip therapies

Liselotte Timp[1]*, Marnix Franken, Manuel AFV Gonçalves, Aurelie Goyenvalle, Anne Bigot, Vincent Mouly, WWM Pim Pijnappel, Jessica C de Greef, Silvère M van der Maarel, Erik H Niks, Remko Goossens, Annemieke M Aartsma-Rus*
[1]LUMC department of Human Genetics, Leiden, Netherlands

2.100P Circulating miRNA biomarkers for Duchenne muscular dystrophy patients with neurodevelopmental disorders

Pak On Patrick Yee[1]*, Martin Cheung, Sophelia Hoi-Shan Chan*
[1]Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong, China

2.101P Role of transient receptor potential melastatin 7 (TRPM7) in the pathogenesis of a Duchenne muscular dystrophy mouse model

Takuya Yokoyama[1,2]*, Kotaro Hirano, Masayuki Matsushita, Yasuo Mori, Yuji Hara*
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2.102P Brain microstructural alterations in patients with Duchenne muscular dystrophy: a diffusion kurtosis imaging study

Pak On Patrick Yee[1]*, Hui Zhang, Chengzhe Zhang, Richard Ng, Ka Fung Henry Mak, Tatia Mei Chun Lee, Hanns Lochmüller, Sophelia Hoi-Shan Chan*
[1]Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong, China

2.103P Genotype–phenotype correlations across motor, cognitive, and cardiac outcomes in a large dystrophinopathy cohort

Seungjae Lee[1]*, Seungbok Lee, Eun Na Kim, Byung Chan Lim, Anna Cho, Jong-Hee Chae, Young Ho Kim*
[1]Seoul National University Children's Hospital, Seoul, South Korea

2.104P Muscle-on-a-chip models for characterization of dystrophin-dependent transcriptional signalling disruptions in Duchenne muscular dystrophy

Remko Goossens[1,2]*, Liselotte Timp, Pim Pijnappel, Silvère van der Maarel, Annemieke Aartsma-Rus*
[1]Leiden University Medical Center, Leiden, Netherlands, [2]Duchenne Center Netherlands, Leiden/Nijmegen/Heeze, Netherlands

2.105P Exploring the function and localization of the dystrophin short isoform Dp140 in the brain
Yuki Ashida[1], *Hironaka Igarashi, Hiroshi Kuniishi, Takahiro Fujimoto, Tadashi Okamura, Eri Takeuchi, Masayuki Sekiguchi, Norio Motohashi, Yoshitsugu Aoki*
[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

Poster Session 2: 2.073eP-2.085eP, 2.106P-2.128P: Pathophysiology of NMDs

2.073eP Leveraging patient-derived myoblasts and FAPs to study neuromuscular diseases
Anne Bigot[1], *Kamel Mamchaoui, Jessica Ohana, Lylia Mekzine, Mona Bensalah, Ngoc Dong Pham Phan, Gillian Butler-Browne, Elisa Negroni, Capucine Trollet, Vincent Mouly*
[1]*Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris, France*

2.074eP Acute H2O2 exposure remodels chromatin accessibility in skeletal muscle stem cells toward differentiation-associated regulatory programs
Lyn Eigenfeldt[1,2], *Tongta Sae-Ong, Christopher Nelke, Tobias Ruck*
[1]*University Hospital Düsseldorf, Düsseldorf, Germany,* [2]*Heimer Institute for Muscle Research, Bochum, Germany*

2.075eP p21 contributes to senescent myoblast persistence and impaired regeneration in age-related sarcopenia
Sujin KIM, ***Sohee Moon***[1,2], *Chan-Young So, Dwifrista Vani Pali, Keren Esther Kristina Mantik, Ju-Hee Kang*
[1]*Department of Pharmacology, College of Medicine, Inha University, Incheon, South Korea,* [2]*Research Center for Controlling Intercellular Communication, College of Medicine, Inha University, Incheon, South Korea*

2.076eP CCL5, a candidate gene associated with age-related sarcopenia, attenuates palmitic acid-induced myotube atrophy
Chan-young So, ***Dwifrista Vani Pali***[1,2,3], *sujin KIM, Keren Esther Christina Mantik, Sohee Moon, Ju-Hee Kang*
[1]*Department of Pharmacology, College of Medicine, Inha University, Incheon, South Korea,* [2]*Research Center for Controlling Intercellular Communication, College of Medicine, Incheon, South Korea,* [3]*Program in Biomedical Science and Engineering, Inha University, Incheon, South Korea*

2.077eP N-acetylglucosamine induces coordinated flow-like migration of myoblasts underlying efficient myogenesis
Masahiko Sato[1], *Sachiko Sato, Ann Rancourt, Guillaume St-Pierre*
[1]*Centre de recherche du CHU de Québec-Université Laval, Quebec City, Canada*

2.078eP Systematic evaluation of myotube formation and pepsin concentration dependency of plant-derived protein hydrolysates using C2C12 cells
Ryohei Sakuraoka[1], *Hirofumi Masutomi, Katsuyuki Ishihara*
[1]*Calbee Inc., Utsunomiya, Tochigi, Japan*

2.079eP Asymmetric nucleocytoplasmic transport of desmin: lack of karyopherin-independent import, and evidence for hydrophobicity-driven export
Pervin Dincer[1], *Ecem Kural Mangıt*
[1]*Hacettepe University Medical Biology, Ankara, Turkey*

2.080eP Increased transient receptor potential melastatin 2 expression in a mouse model of inflammatory muscle pain
Mayuu Iwai[1], *Yui Fukushima, Mei Yamada, Angela F. Smith, Kathleen A. Sluka, Rina Okura, Yuri Okada, Naoki Ooka, Kenshiro Kashima, Momoko Nagai-Tanima, Akira Ito, Shigehito Yamada, Kazuhiro Hayashi*
[1]*Human Health Sciences, Graduate School of Medicine, Kyoto University, Kyoto, Japan*

2.081eP Glutamine enhances myotube formation in C2C12 myoblasts through glutamine metabolism-mediated histone H3 acetylation that elevates myogenin gene expression
Masaru Takatoya[1,2], *Daichi Arai, Tomoya Kasugai, Tsukasa Tominari, Yoshitsugu Aoki, Masaki Inada*
[1]*Department Of Molecular Therapy, National Institute Of Neuroscience, National Center Of Neurology And Psychiatry, Tokyo, Japan,* [2]*Department of Biotechnology and Life Science, Tokyo University of Agriculture and Technology, Tokyo, Japan*

2.082eP ANTXRopathies: a unified spectrum of ANTRX1 and ANTRX2 disorders driven by extracellular matrix dysfunction
Dipti Baskar[1], *Atchayaram Nalini, Seena Vengalil, Gautham Arunachal, Ananthapadmanabha Kotambail, Shilpa Rao*
[1]*National Institute of Mental Health and Neuro Sciences (NIMHANS), Bengaluru, India, Bengaluru, India*

2.083eP Machine learning-directed exercise protocol development for 3D neuromuscular junction tissues
Nelsa Estrella[1], *Jacob Fleming, Thomas Leahy, Hamed Ghazizadeh, Christal Worthen, Greg Luerman, Nicholas Geisse*
[1]*Curi Bio, Seattle, United States*

2.084eP The preventive effects of mild hyperbaric oxygen environment on age-related sarcopenia through integrated regulation of metabolism and capillary vascularization
Juzhen Chen[1], *Jihao Xing, Tiancheng Tang, Zhifan Liu, Noriaki Maeshige, Hidemi Fujino*
[1]*Kobe University Graduate School Of Medicine, Kobe, Japan*

2.085eP. GLP-1 receptor agonist therapy in obesity with Duchenne muscular dystrophy: a case highlighting risk-benefit tradeoffs
Hannah Walsh, Ruby Joshi Batajoo, Danielle Sharon, Audrey McKennitt, Jillian Belgrad, ***Courtney Shellenbarger***[1], *Rebecca Artinian, Celina Goncalves, Brenda Wong, Marla Mungovan, Stephen Chrzanowski*
[1]*UMass Memorial Medical Center, United States*

2.106P Dystrophin controls clathrin-mediated endocytosis and endolysosomal homeostasis
Eline Lemerle[1], *Simon Giraud, Satish Babu Moparthi, Marion Benoist, Jeanne Lainé, Quentin Miagoux, Kamel Mamchaoui, Beatrice Labella, Teresinha Evangelista, Xavier Nissan, Helge Amthor, Luis Garcia, Stéphane Vassilopoulos*
[1]*Inserm, Institute of Myology, Paris, France*

2.108P No evidence of the presence of dystrophin protein in rodent muscle stem cells
Meriem Matouk, Adrien Morin, Gaspard Macaux, Brendan Evano, Pascal Maire, Vincent Mirouse, Sestina Falcone, ***Helge Amthor***[1]
[1]*Unité de Recherche PHARMAColigo UR20261, Université de Versailles Saint-Quentin-en-Yvelines, Versailles, France*

2.109P Clinical and molecular expansion of SNUPN disease: proteomics reveals ECM remodeling and mitochondrial dysfunction
Göknur Haliloğlu[1], *Sebahattin Çırak, Andreas Hentschel, Heike Kölbel, Anne Schänzer, Andreas Roos*
[1]*Hacettepe University, Faculty of Medicine, Department of Pediatrics, Division of Pediatric Neurology, Ankara, Turkey*

2.110P Bee venom modulates skeletal muscle regeneration and inflammatory responses
N Bergachi, ***Emna Farhat***[1], *C Ellijimi, A Krir, R Ben Khalifa, S Ben Dahou, B Constantin, A Cheikh*
[1]*The Muscle Lab. Iris Medical Centre, Hedi Nouira Avenue, Ennasr2, 2037., Tunis, Tunisia*

2.111P Hypobaric hypoxia-induced miR-1 expression regulates myogenesis by modulating Pax3 and Hsp70 signaling pathway in skeletal muscle
Samrita Mondal[1], *Richa Rathor, Geetha Suryakumar*
[1]*Defence Institute of Physiology and Allied Sciences (DIPAS), DRDO, Delhi, India*

2.112P Multi-layered nitration of HGF/c-met axis impairs muscle regeneration during aging
Alaa Magdi Hassanein Elgaabari[1,2], *Junri Miyamoto, Kahona Zushi, Mako Nakamura, Takahiro Suzuki, Ryuichi Tatsumi*
[1]*Kafrelsheikh University, Kafrelsheikh, Egypt,* [2]*Kyushu University, Fukuoka, Japan*

2.113P Primary cilium orchestrates human muscle stem cell differentiation though Indian hedgehog-mediated signaling
Ana Saric[1], *Lola Albertella, Olivier Conscience, Paola Frankovic, Baptiste Periou, Sarah Souvannanorath, Frédéric Relaix, Edoardo Malfatti, Ludovic Martin, François Jérôme Authier*
[1]*Paris Est Creteil University, INSERM, IMRB U955-Team MUSKETHER, Physiopathology and Therapy of Musculoskeletal System Diseases, Creteil, France*

2.114P Combinatorial treatment of anti-oxidant NAC and antisense oligonucleotides can improve treatment effect in mdx mice by improved dystrophin distribution and reduced pathology
Anne-Fleur Schneider[1], *Galina Filonova, Christa Tanganyika-de Winter, Maaike van Putten, Annemieke Aartsma-Rus, Remko Goossens*
[1]*Leiden University Medical Centre, Leiden, Netherlands*

2.115P Elucidating drivers of cardiac remodelling in Duchenne muscular dystrophy
Elisa Villalobos[1,2], *Rachel Redgrave, James Clark, Guy MacGowan, Alberto Malerba, Richard Piercy, Gavin Richardson, Francesco Muntoni*
[1]*Dubowitz Neuromuscular Centre, Great Ormond Street Institute of Child Health, University College London, London, United Kingdom,* [2]*Translational and Clinical Research Institute, Faculty of Medical Sciences, Newcastle University, Newcastle Upon Tyne, United Kingdom*

2.116P Fibro-adipogenic progenitors enhance human 3D tissue engineered skeletal muscles: a platform for muscular dystrophy research
Roy Augustinus[1], *Lotte de Ridder, Dongxu Zheng, Marnix Franken, Judit Balog, Patrick van der Vliet, Alessandro Iuliano, Remko Goossens, Johanna Hamel, Pim Pijnappel, Jessica de Greef, Silvere van der Maarel*
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2.117P MyoPath: a deep learning pipeline for objective morphometric assessment of skeletal muscle biopsies
Huahua Zhong[1], *Mingshi Gao, Shang Ma, Wenhan Zhang, Nachuan Chen, Kexin Jiao, Bochen Zhu, Jie Song, Chong Yan, Dongyue Yue, Jianying Xi, Zunguo Du, Wenhua Zhu, Chongbo Zhao, Sushan Luo*
[1]*Huashan Hospital, Shanghai, China*

2.118P Unlocking muscular dystrophies using patients' induced pluripotent stem cells
Imane Ben Daoud[1], *Mégane Delourme, Raphaël Chevalier, Defne Aksoy, Fabio Neto Da Silva, Charlotte Vachet, Jérôme Robin, Emmanuelle Salort-Campana, Frédérique Magdinier, Leslie Caron*
[1]*Marseille Medical Genetics, Aix-Marseille University, INSERM, Marseille, France*

2.119P Genetic characterisation of SLONM: insights from nine biopsy-confirmed cases
Jana Marie Schwarz[1], *Felix Kleeefeld, Johannes Prudlo, Andreas Roos, Werner Stenzel*
[1]*Charité - Universitätsmedizin Berlin, Berlin, Germany*

2.120P Molecular mechanisms of skeletal muscle regeneration mediated by mechanosensitive ion channels
Yuji Hara[1], *Kotaro Hirano*
[1]*University of Shizuoka, Shizuoka, Japan*

2.121P Functionally informed screening of gene therapy capsid transduction efficiency using human 3D engineered muscle tissues
Thomas Leahy, ***Nelsa Estrella***[1], *Travis Moerk, Jacob Fleming, Kevin Gray, Christal Worthen, Greg Luerman, Nicholas Geisse*
[1]*Curi Bio, North Andover, United States*

2.122P Spatial transcriptomic profiling reveals compensatory and degenerative pathways in VCP-MSP muscle across histological severity
Marianela Schiava[1,2], *Sandra de Haan, Pietro Spitali, Yolande Parkhurst, Dan Cox, Andrew Galloway, James Clark, Jordi Diaz Manera*
[1]*The John Walton Muscular Dystrophy Research Centre and Newcastle Hospitals NHS Foundation Trusts, Newcastle Upon Tyne, United Kingdom,* [2]*Newcastle Hospitals NHS Foundation Trusts, Newcastle upon Tyne, United Kingdom*

2.123P Collagen VI is a fibrosis-associated signal disrupting muscle regeneration across distinct human myopathies
Elisa Melissa Negroni[1], *Laura Muraine, Mona Bensalah, Stephen Gargan, Paul Dowling, Anne Bigot, Valérie Allamand, Jamila Dhiab, Maria Kondili, Sophie Perié, Jean Lacau St-Guily, Gillian Butler-Browne, Vincent Mouly, Kay Ohlendieck, Capucine Trollet*
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2.124P Dynamics of muscle stem/niche cells in regeneration and disease
Shahragim Tajbakhsh[1]
[1]*Institut Pasteur Paris, Paris, France*

2.125P S-nitrosylation at Cys620 establishes a redox–phase separation axis modulating androgen receptor dysfunction in SBMA
Yasumasa Hashimoto[1,2,3], *Antonio Garcia-Guerra, Kseniya Korobchevskaya, Divya Seth, Ka Long Ko, Taichi Fukunaga, Marco Fritzsche, Richard Premont, Jonathan S. Stamler, Matthew J A Wood, Carlo Rinaldi*
[1]*Department of Neurology, Kansai Medical University, Osaka, Japan,* [2]*Institute of Developmental and Regenerative Medicine (IDRM), University of Oxford, Oxford, United Kingdom,* [3]*Department of Paediatrics, University of Oxford, Oxford, United Kingdom*

2.127P Structural and recombination-related determinants of CMT1A locus instability across populations
Pedro M Rodriguez Cruz[1,2,3], *Timofey Prodanov, Alexandra Alitsiou, Henriette Senghor, Anne-Sophie Lia-Baldini, Karima Ghorab, Amadou Gallo Diop, Moustapha Ndiaye, Sergi Beltran, Oscar Lao*
[1]*Centro Nacional De Analisis Genómico, Dakar, Senegal,* [2]*Universite Cheikh Anta Diop, Dakar, Senegal,* [3]*Neurology Department, Centre Hospitaliere National Universitaire de Fann, Dakar, Senegal*

2.128P Novel tripartite neuromuscular junction model via direct reprogramming of human urine-derived cellsfor elucidating amyotrophic lateral sclerosis pathogenesis
Kazuki Okoshi[1,2], *Katsuhiko Kunitake, Norio Motohashi, Yoshitsugu Aoki*
[1]*National Center for Neurology and Phychiatry, Kodaira Tokyo, Japan,* [2]*Institute of Science Tokyo, Bunkyo Tokyo, Japan*

Poster Session 2: 2.086eP-2.090eP: Environmental Sustainability

2.086eP Eco-friendly, home-based rehabilitation using calibrated DIY equipment for LGMD (R1-R5) and GNE myopathy in India: a prospective, randomized, open-label, blinded endpoint trial

Sneha Sneha[1], *Nimita Rani, Zubina Khan, Ajay Garg, Ayush Agarwal, Hina Narzari, Nilima Nilima, Rohit Bhatia, Venugopalan Vishnu*
[1]*Department of Neurology, All India Institute of Medical Sciences, New Delhi, India*

2.087eP Printed versus electronic posters: Environmental footprint and other considerations

Jaakko Sarparanta[1,2,3], *WMS Environmental Sustainability Committee*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Helsinki, Finland,* [3]*WMS Environmental Sustainability Committee, London, United Kingdom*

2.088eP PHARMA solidaires, a circular social model for medical devices and parapharmaceutical redistribution : five-year sustainability data

Sami Tayb-Boulahfa[1,2], *Catherine Genton, Solène Emedy, Guillaume Hébert, Aurélie Deltombe, Aurélie Mayeux, Julia Aubé, Aline Lazberg*
[1]*Centre de Recherche en Myologie, UMRS 974, INSERM-Sorbonne University, Paris, France,* [2]*PHARMA solidaires organisation, Paris, France*

2.089eP The lipid paradox in sarcopenia: a nationwide mediation analysis of the role of body volume loss in muscle wasting (KNHANES 2022–2024)

Jung Hwan Lee[1]
[1]*Seoul St. Mary's Hospital, College of Medicine, the Catholic University of Korea, Seoul, South Korea*

2.090eP Multidisciplinary approach to weight management and nutrition in genetic neuromuscular diseases

Farah J Walter, Simone Thiele, Gert Bischoff, Monika Bischoff, Eva Malm, Caterina Wendel, Daniela Grote, Stephan Wenninger, ***Maggie Walter***[1]
[1]*Friedrich-Baur-Institute, Dept. Of Neurology, University Hospital, Lmu Munich, Munich, Germany*

Short Oral Presentations 4: SP16-SP20, Congenital myopathies

SP16 MYH7B biallelic variants cause proximal and axial myopathy with respiratory insufficiency

Diana Laure Mboumba, Marion Onnée, Valentina Taglietti, Salla M Kangas, Reetta Hinttala, Johanna Uusimaa, Aurelio Hernandez-Lain, Cristina Dominguez-Gonzalez, Abdelrakaz Aissat, Baiba Lāce, Inna Inashkina, Pawel Zayakin, Pascal Fanen, Alix de Becdelièvre, ***Edoardo Malfatti***[1,2]
[1]*Univ Paris-Est Créteil, INSERM, U955 IMRB, 94010 , Crétéil, France,* [2]*APHP, Centre de Référence des Maladies Neuromusculaires, Hôpital Universitaire Henri Mondor, Créteil, France, Creteil, France*

SP17 A familial congenital myopathy supports a muscle-specific biallelic coding-regulatory mechanism in IARS1

Adrien Rihoux[1,2]*, Sébastien Audet, Alexie Gagné, Gionvani Risato, Vivian Thai, Clémence Gonthier-Cummings, Jennifer Hautecloque, Audrey Labarre, Alex Parker, Erin O'Ferrall, Jason Karamchandani, Kessen Patten, Bernard Brais, Martine Tétreault*
[1]*University of Montreal, Montreal, Canada,* [2]*CHUM Research Center, Montreal, Canada*

SP18 A natural history study to define clinical outcomes and trial readiness in STAC3 myopathy

Eleonora D'Ambrosio[1]*, Danielle Kokoski, Christopher Hemond, Katherine Luzuriaga, Adrian Zai, Robert H Brown*
[1]*UMass Chan Medical School, Worcester, United States*

SP19 Genotype-driven classification of cardiac phenotypes in patients with biallelic titinopathies: a retrospective study on 84 patients

Maria Francesca Di Feo[1,2,3]*, Heinz Jungbluth, Jennifer Roggenbuck, TTN biallelic cardiomyopathy study group, Bjarne Udd, Job A J Verdonschot, Marco Savarese*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Helsinki, Finland,* [3]*University of Genoa, Genoa, Italy*

SP20 A high-content cell painting platform identifies a morphological disease signature for drug discovery in SELENON-congenital myopathy cell models

Pamela Barraza Flores[1]*, Shatavisha Dasgupta, Seth Thor, Won Lee, Beth Cimini, Maria Alimova, Alan Beggs*
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Short Oral Presentations 5: SP21-SP24, Myotonic dystrophy mechanisms

SP21 The genetic landscape of DM1: ancestry-specific and phenotypic insights from one million genomes

Rezbieara Rahman[1]*, Marta Futema, Elijah Behr, Emma Matthews*
[1]*Neuroscience and Cell Biology Research Institute, School of Health & Medical Sciences, City St George's, University of London, London, United Kingdom*

SP22 DM1-Hub: integrative population-scale long-read genomics to resolve locus-specific and genome-wide modifiers in myotonic dystrophy type 1

Alvaro S Larran Mottino[1]*, Marc Corral-Juan, Luis Orduña Rubio, Daniel M. Borràs, Raquel Pérez Gómez, Eva Coll Liesa, Alicia López-Martín, Eric Casellas Sanfelix, Alejandro Montalva-Gimenez, Anna Revert-Barberà, Clara Moliner Diaz, Alba Herrero Gómez, Gerardo Gutiérrez-Gutiérrez, Andone Sistiaga, Jorge Alonso-Pérez, Sebastián Figueroa-Bonaparte, Virginia Arechavala-Gomeza, Arturo López-Castel, Gisela Nogales Gadea, DM1-Hub Consortium*
[1]*Germans Trias i Pujol Research Institute (igtp), Badalona, Spain*

SP23 Ultrastructural alterations in DM1 muscle cells revealed by cryo soft x-ray tomography

Pau Maestre-Mora[1]*, Judit Nuñez-Manchón, Eva Coll Liesa, Alba Herrero-Gómez, Beatriz Llamusi, Estefanía Cerro-Herreros, Mònica Suelves, Ana Joaquina Pérez-Berná, Gisela Nogales-Gadea*
[1]*Badalona Neuromuscular Research Group (GRENBA), Germans Trias i Pujol Research Institute (IGTP), Badalona, Spain*

SP24 GDF-15 and FGF-21 in myotonic dystrophy type 2: linking mitochondrial dysfunction to clinical severity and multimodal outcomes

Alexander Mensch, Stojan Peric, Andreas Roos, Nicolina Südkamp, Tobias Ruck, ***Felix Kleefeld***[1]
[1]*Bg Universitätsklinikum Bergmannsheil Bochum, Bochum, Germany*

Short Oral Presentations 6: SP25-SP27, Modeling disease mechanisms

SP25 Generation and characterization of facioscapulohumeral muscular dystrophy model mice carrying an artificial chromosome loaded with a 5 Mb chromosome 4q35

Yosuke Hiramuki[1]*, Miwa Hosokawa, Ichizo Nishino, Hiroyuki Kugoh, Yasuhiro Kazuki*
[1]*Department of Chromosome Biomedical Engineering, Tottori University, Yonago, Japan*

SP26 MyoScore: a genetically informed transcriptomic scoring system for quantifying human skeletal muscle health

Huahua Zhong[1], *Shang Ma, Victoria Lillback, Mingshi Gao, Wenhan Zhang, Mridul Johari, Ali Oghabian, Per Harald Jonson, Jianying Xi, Wenhua Zhu, Shanfeng Zhu, Peter Hackman, Bjarne Udd, Chongbo Zhao, Marco Savarese, Sushan Luo*

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SP27 Platforms enabling reclassification of VUS in neuromuscular and neurogenetic diseases

Gina Ravenscroft[1], *Jevin Parmar, Chiara Folland, Jasmine Chew, Carolin Scriba, Christina Vo, Shelby Mullin, Phillipa Lamont, Margit Shah, Esther Tantsis, Martin Delatycki, Matthew Lynch, Tarin Richie, Daniella Hock, Jozef Gecz, Lachlan Jolly, David Stroud*

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Poster Session 3: 3.001eP-3.005eP, 3.001P-3.012P: Distal myopathies and OPMD

3.001eP Physical functional status of GNE myopathy by self-reported north star assessment for limb girdle muscular dystrophy
Yeonjung Mun[1], *Dae-Seong Kim, JinHong Shin*
[1]*Pusan National University Of Yangsan Hospital, Yangsan, South Korea*

3.002eP Comparative analysis of differential diagnostic features in distal myopathies and distal hereditary motor neuropathies
Dmitrii Subbotin[1], *Artem Borovikov, Olga Shchagina, Oxana Ryzhkova, Nikolay Zernov, Aysylu Murtazina*
[1]*Research Centre for Medical Genetics, Moscow, Russia*

3.003eP Interindividual variability of muscle involvement in familial FLNC-related myopathy on imaging
Wakako Yoshioka[1], *Satoru Noguchi, Hotake Takizawa, Rui Shimazaki, Kayoko Eguchi, Yuji Takahashi, Shinichiro Hayashi, Ichizo Nishino*
[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

3.004eP Intrafamilial phenotypic variability in a Korean family with TTN-related myopathy
Soohyun Kim[1], *Hyung Jun Park, Hyun-Ho Kim*
[1]*Department of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine, Seoul, South Korea*

3.005eP A 47-year-old woman with sporadic late-onset nemaline myopathy superimposed complicating multiple sclerosis: a case report
Fumiko Furukawa[1], *Hotake Takizawa, Toru Yamakawa, Atsuko Tsuneyama, Atsuko Katsumoto, Yasushi Oya, Tomoko Okamoto, Takashi Yamamura, Ichizo Nishino, Yuji Takahashi*
[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

3.001P One-year oral aceneuraminic acid treatment in four patients with GNE myopathy
Nozomu Tawara[1], *Takashi Ueyama, Chikako Nagatoshi, Akie Tawara, Akiko Fujimoto, Masatoshi Ishizaki, Ryoichi Kurisaki, Yasuto Nishida, Yasushi Maeda*
[1]*National Hospital Organization Kumamoto Saishun Medical Center, Kumamoto, Japan*

3.002P SUMOylation studies on SMPX
Jaakko Sarparanta[1,2], *Per Harald Jonson, Helena Luque, Emma James, Charlotte Walder, Kalle Kettunen, Marco Savarese, Bjarne Udd*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Helsinki, Finland*

3.003P Evaluation/characterization of patient-derived MYH7-mutant myogenic progenitor cells as 2D and 3D models of muscle disease
Jeremy Garcia[1], *Joshua Clayton, Anna Johnstone, Nigel Laing, Gianina Ravenscroft, Rhonda Taylor, Peter Houweling, Leon Kiriaev, Vanessa Crossman*
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3.004P Update on the description of 50 patients with p.Ser55Phe variant in the MYOT gene: MYOT-MUR study
Rafael Jenaro Martínez Marín[1], *Gerardo Ricardo Zmork Martínez, Marina Aledo Serrano, Perez Fernández Marta, Perez Lucas Josefa*
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3.005P Developing a miRNA-based single-construct 'knockdown and replace' gene therapy for desminopathy
Andrew Galloway[1], *Emma Friar, Stefan Nicolau, Volker Straub, Scott Harper, Kevin Flanigan, Liubov Gushchina, Michael Keogh*
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3.006P Clinical and genetic features of oculopharyngeal muscular dystrophy in a Chinese cohort
Xinyu Chen[1,2,3], *Kexin Jiao, Chongbo Zhao, Wenhua Zhu, Jianying Xi*
[1]*Huashan Hospital, Fudan University, Shanghai, China,* [2]*National Center for Neurological Disorders (NCND), Shanghai, China,* [3]*Department of Neurology and Rare Disease Center, Shanghai, China*

3.007P Scapular winging is a marker of clinical severity in OPMD: insights from two French-Canadian cohorts
Mayra Aldecoa[1], *Olivia Azancot, Pablo Iruzubieta, Ariane Saint-Denis, Marie Josee Dicaire, Felipe Villa Tobon, Bernard Brais*
[1]*Mcgill University, Montreal, Canada*

3.008P Clinical characteristics and genotype-phenotype correlation of oculopharyngodistal myopathy: a single-center study in China

Kexin Jiao[1], *Xinyu Chen, Dongyue Yue, Mingshi Gao, Chongbo Zhao, Jianying Xi, Wenhua Zhu*
[1]*Department of Neurology and Rare Disease Center, Huashan Hospital, Fudan University, Shanghai, and National Center for Neurological Disorders (NCND), Shanghai, China*

3.009P A study of Fibronectin and Periostin in GNE-related myopathy unravels new biomarkers of pathophysiological relevance

Andreas Roos[1], *Corinna Preusse, Alexa Derksen, Kristl Claeys, Andreas Hentschel, Benedikt Schoser, Ulrike Schara-Schmidt, Mira Schiefer, Vera Dobelmann, Lyn Eigenfeldt, Sally Spendiff, Teresinha Evangelista, Oksana Pogoryelova, Marco Savarese, Hanns Lochmüller, Tobias Ruck*
[1]*University Medicine Essen, Dept. of Pediatric Neurology, Essen, Germany*

3.010P FLNC-related myopathy: a description of 27 unrelated patients from 26 families

Manh-Louis Nguyen[1,2], *Beatrice Labella, Teresinha Evangelista, Nathalie Streichenberger, Gisèle Bonne, Pascale Richard, Karim Wahbi, Philippe Petiot, Beatrice Lannes, Aleksandra Nadaj-Pakleza, Marie-Laure Martin-Négrier, Guilhem Sole, Pascale Marcorelles, Yann Pereon, Jean Baptiste Noury, Thierry Maisonobe, Corinne Métay, Anthony Béhin, Tanya Stojkovic*
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3.011P Cure MFM13: a patient-driven charitable initiative advancing research in MFM13

Sylwia Szwec[1], *Anna Kordala, Matt McLeod, Julia Mielcarz, Todd King*
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3.012P Engineering 3D human skeletal muscle models to explore fibrosis in the oculopharyngeal muscular dystrophy

Maria Bergas Buades[1], *Laura Muraine, Mona Bensalah, Jessica Ohana, Kamel Mamchaoui, Valentine Hanique, Nami Altin, Penney Gilbert, Stéphanie Descroix, Giacomo Gropplero, Jean Lacau St-Guily, Gillian Butler-Browne, Vincent Mouly, Anne Bigot, Capucine Trollet, Elisa Negroni*
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Poster session 3: 3.006eP-3.015eP, 3.013P-3.020P: Drug-induced and infectious myopathies

3.006eP Phenotypic heterogeneity and clinical characterisation of SSRI-induced lipid storage myopathy: a case series

Joshua Winderlich[1], *Catriona McLean, Edrich Rodrigues*
[1]*Alfred Health, Melbourne, Australia*

3.007eP Colchicine myopathy: early recognition through myoedema sign and electrodiagnostic findings

Hung Dang[1]
[1]*University Medical Center Ho Chi Minh City, Ho Chi Minh City, Vietnam*

3.008eP Disruption of the mitochondria–microtubule axis as mechanism of monensin-induced myotoxicity and therapeutic potential of arginine

Xiaoqing Lyu[1], *Yifei Feng, Pengfei Lin, Chuanzhu Yan*
[1]*Qilu Hospital of Shandong University, Jinan, China*

3.009eP Spatial dynamics between resident immune cells involve an IFN and IL15 axis in immune checkpoint blockade-induced muscle toxicity

Julian Sanchez-Dal Cin[1], *Céline Anquetil, Sarah Léonard-Louis, Joe-Elie Salem, Damien Amelin, Bérénice Tendrel, Aude Rigolet, Nicolas Champtiaux, Pascale Daniel, Nadege Wesner, Michelle Rosenzwajg, Fathia Mami-Chouaib, Olivier Benveniste, Stéphanie Corgnac, Yves Allenbach*
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3.010eP Increased incidence of idiopathic inflammatory myopathies after the SARS-CoV-2 outbreak: an interrupted time-series study from the French national health data system and a myositis-specific autoantibody database

Dylan Vellas[1,2], *Audrey Bergès, Yann De Rycke, Sandrine Pinto, Antoine Rozes, Olivier Benveniste, Florence Tubach, Yves Allenbach*
[1]*Department of Internal Medicine and Clinical Immunology, Inflammatory Myopathies Reference Center, Pitié-Salpêtrière hospital, Greater Paris University Hospitals, Paris, France,* [2]*Sorbonne Université, INSERM, Institut Pierre Louis d'Epidémiologie et de Santé Publique, Equipe PEPITES, AP-HP, Hôpital Pitié-Salpêtrière, Département de Santé Publique, Centre de Pharmacoépidémiologie (Cephepi), CIC-1901, F75013, Paris, France*

3.011eP Three cases of acquired lysosomal myopathies: a case series and literature review
Tingjun Dai[1,2,3], *Xinyi Gao, Guoyong Zhang, Wei Zhang, Chuanzhu Yan*
[1]*Department of Neurology, Qilu Hospital of Shandong University, Jinan, China,* [2]*Shandong Key Laboratory of Mitochondrial Medicine and Rare Diseases, Jinan, China,* [3]*Research Institute of Neuromuscular and Neurodegenerative Diseases, Jinan, China*

3.012eP Clozapine-induced toxic Myopathy and rhabdomyolysis: evidence from proteomic profilinm
Beatrice Labella[1,2], *Emmanuelle Lacene, Anaïs Chanut, Guillaume Le Maistre, Sara Walli, Tobias Ruck, Anastasia Alexandridou, Andreas Roos, Teresinha Evangelista*
[1]*Pitié-Salpêtrière Hospital, Paris, France,* [2]*Myology Institute, Paris, France*

3.013eP Hydroxychloroquine skeletal muscle toxicity in a referral biopsy cohort: light microscopic and ultrastructural features
Osorio Lopes Abath Neto[1], *Erica Fossee, Melissa Mejia-Bautista, Kathryn Eschbacher, Steven Moore*
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3.014eP Severe Colchicine-induced neuromyopathy mimicking acute surgical abdomen with prominent subclinical myotonic discharges: A case report
Sani Tavivatana[1], *Nattharadee Phutthinart, Theerawat Kumutpongpanich, Jantima Tanboon*
[1]*Department of Medicine, Sirindhorn Hospital, Bangkok Metropolitan Administration, Bangkok, Thailand*

3.015eP Intramyofiber leukemic infiltration as an isolated extramedullary relapse of acute myeloid leukemia with monocytic differentiation
Tingjun Dai[1,2,3], *Xiaoyu Liu, Chuanzhu Yan*
[1]*Qilu Hospital Of Shandong University, Jinan, China,* [2]*Shandong Key Laboratory of Mitochondrial Medicine and Rare Diseases, Jinan, China,* [3]*Research Institute of Neuromuscular and Neurodegenerative Diseases, Jinan, China*

3.013P Metagenomics for pathogen identification in inflammatory myopathies: a prospective observational study
Yves Allenbach[1], *Andrei-Valentin Brîncuș, Cécile Angebault, Nadège Wesner, Sarah Leonard-Louis, Alexandre Le Joncour, Camille Blandin, Pascal Richette, Philippe Poirier, Olivier Benveniste, Christophe Rodriguez, Françoise Botterel*
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3.014P Evidence for transcriptomic changes impacting muscle contraction in patients with post-acute COVID-19 syndrome
Gianmarco Severa, Rakel Altabe-Mellul, Alessio Rotini, Peggy Lafuste, Sarah Souvannanorath, Edoardo Malfatti, Emmanuel Itti, Hakim Ahmed-Belkacem, Fred Relaix, ***Francois Jerome Authier***[1]
[1]*Paris Est-creteil University, Creteil, France*

3.015P Aluminium hydroxide-induced macrophagic myofasciitis (MMF)
Mehdi Aoun Sebaiti[1], *Guillemette Crepeaux, Jean-Daniel Masson, Baptiste Periou, Romain K Gherardi, Emmanuel Itti, Francois Jerome Authier*
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3.016P Sertraline-associated MADD is an emerging riboflavin-responsive lipid storage myopathy with mitochondrial changes – a nationwide population-based study
Ulrika Lindgren[1], *Sofie Sunebo, Emil Nilsson, Martin Engvall, Carola Hedberg-Oldfors, Yan Shen, Inger Nennesmo, Olof Danielsson, Anders Oldfors, Hanna Appelqvist*
[1]*University of Gothenburg / Sahlgrenska University Hospital, Gothenburg, Sweden*

3.017P Sertraline-induced toxic myopathy: Clinical course, treatment response, pathomorphology and first lipidomic insights
Willem De Ridder, Karsten Krause, Belal Alshaar, Heidrun Krämer, Tobias Ruck, Sven Heiles, Jonathan Baets, Anne Schänzer, ***Andreas Roos***[1]
[1]*University Medicine Essen, Dept. of Pediatric Neurology, Essen, Germany*

3.018P Histomorphologic and multi-omics characterization of immune checkpoint inhibitor–associated neuritis
Corinna Preusse[1], *Maria Casal-Dominguez, Marie-Therese Holzer, Andreas Roos, Andreas Hentschel, Hans-Christian Koennecke, Leonie Müller-Jensen, Iago Pinal-Fernandez, Andrew Mammen, Anne Schänzer, Werner Stenzel*
[1]*Charité – Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt-Universität zu Berlin, Department of Neuropathology, Charitéplatz 1, 10117 Berlin, Germany, Berlin, Germany*

3.019P Late-onset multiple Acyl-CoA dehydrogenase deficiency: clinical, pathological, and genetic findings in a Brazilian cohort
Ana Luisa Hernandez[1], *Andre Macedo-Silva, Mariana Hiromi Manoel Oku, Cristiane de Araújo Martins Moreno, José Pedro Baima, Eliene Dutra Campos, Rodrigo de Holanda Mendonça, José Luiz Pedroso, Alzira Alves de Siqueira Carvalho, Edmar Zanoteli*
[1]*HCFMUSP, São Paulo, Brazil*

3.020P Occurrence of adult-onset lipid storage myopathy in southwestern Sweden: an incidence study

Carola Hedberg-oldfors[1,2,3], *Anders Oldfors, Ulrika Lindgren*

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Poster Session 3: 3.016eP-3.024eP, 3.021P-3.028P: FSHD - Clinical care and case reports

3.016eP De novo facioscapulohumeral muscular dystrophy: a severe subtype driven by D4Z4 repeat contractions

Qifang He, Yuhua Lin, Zeling Zheng, ***Xinrong Xu***[1]*, Zhiqiang Wang*

[1]Department Of Neurology Of First Affiliated Hospital, Fujian Medical University, Fuzhou, China,

3.017eP Accelerated motor function and risk factors in early-onset FSHD1: a retrospective longitudinal cohort study

Xiaodan Lin, Liangliang Qiu, Lin Lin, ***Xinrong Xu***[1]*, Zhiqiang Wang*

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3.018eP Homozygosity for autosomal dominant facioscapulohumeral muscular dystrophy (FSHD)

Ayse Nur Ozdag Acarli, Sahin Avci, ***Ilaha Bakirli***[1]*, Serpil Eraslan, Hulya Kayserili, Piraye Oflazer*

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3.019eP Severe phenotype in early-onset facioscapulohumeral muscular dystrophy: a case report of two siblings

Unsal Yilmaz[1]*, Gamze Gorgulu, Merve Yavuz*

[1]University of Health Sciences Turkey, Izmir Faculty of Medicine, Dr. Behçet Uz Children's Hospital, Izmir, Turkey

3.020eP Comprehensive characterization for Taiwanese patients with fascioscapulohumeral muscular dystrophy

Wen-Chen Liang[1,2,3]*, Chen-Hwa Wang, Chao-Ju Chen, Shyh-Shin Chiou, Yuj-Jyh Jong*

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3.021eP Marked nonpenetrance in a three-generation FSHD1 family despite shared D4Z4 contraction and distal hypomethylation

Yali Hong[1]*, Xinrong Xu, Zhiqiang Wang*

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3.022eP Infantile epileptic spasms syndrome revealing FSHD1 with complex D4Z4 somatic mosaicism

Nicolas Deconinck[1]*, Ophélie Vancutsem, Catheline Vilain, Jean-Paul Kaleeta, Gérald Glibert, Pauline Dontaine*

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3.023eP Unilateral lower extremity atrophy as the sole manifestation of facioscapulohumeral muscular dystrophy type 1 (FSHD-1)

Arya Ebrahimi[1]*, Payam Soltanzadeh, Fatemeh Mofakham, Christopher Brewer*

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3.024eP Co-occurrence of FSHD and myasthenia gravis: findings from the UK FSHD patient registry and the TriNetX global health research network

Lucy Hickson[1]*, Aleks Carver, John McConville, Chiara Marini Bettolo, Gashirai Mbizvo, Gregory Lip, Amy Jayne McKnight, Grace McMacken*

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3.021P Sleep patterns and sleep-related disorder in Indonesian patients with facioscapulohumeral muscular dystrophy: preliminary observations

Dinda Larastika Riyanto[1,2]*, Astri Budikayanti, Adrian Ridski Harsono, Nurul Fadli, Luh Ari Indrawati, Winnugroho Wiratman, Ahmad Yanuar Safri, Fitri Octaviana, Manfaluthy Hakim*

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3.022P Detailed clinical and demographic characteristics of patients with genetically confirmed facioscapulohumeral muscular dystrophy: single-center data from Turkiye

Ayse Nur Ozdag Acarli[1]*, Serpil Eraslan, Ilaha Bakirli, Sahin Avci, Fahrettin Sertaç Yapar, Gulshan Yunisova, Ayfer Arduc Akcay, Mursen Uraz, Arzu Baygul, Ilker Eren, Mehmet Demirhan, Hulya Kayserili, Piraye Oflazer*

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3.023P Natural history and outcome measures in facioscapulohumeral muscular dystrophy in India

Vishnu Venugopalan Yamuna[1], *Nimita Rani, S Sneha, Zubina Khan, Ankur Goyal, Deepti Siddharthan, Devesh Kumawat, Anaha Shibu, Saurabh Mittal, Md Zishan, Ayush Agarwal, Ajay Garg*

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3.024P The natural history of childhood-onset FSHD in an Australian cohort: the iFSHD-LOS study

Katy de Valle[1,2,3], *Chantal Coles, Claire Hogan, Kate Carroll, Peter Houweling, Ian Woodcock*

[1]*The Royal Children's Hospital Melbourne, Department of Physiotherapy (RCH), Melbourne, Australia,* [2]*The Royal Children's Hospital Melbourne, Department of Neurology (RCH), Melbourne, Australia,* [3]*Murdoch Children's Research Institute Melbourne (MCRI), Melbourne, Australia*

3.025P What mimics FSHD? Final diagnoses in patients referred for D4Z4 testing without repeat contraction

Anna Kuchina[1], *Darya Sherstyukova, Mikhail Skoblov, Nikolay Zernov, Dmitrii Subbotin, Artem Borovikov, Elena Dadali, Inna Sharkova, Polina Chausova, Oxana Ryzhkova, Aysylu Murtazina*

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3.026P FSHD-negative phenotypes: diagnostic challenges and clinical implications

Alisha Reyaz[1], *Nimita Rani, Richard J. L. F. Lemmers, Tanveer Ahmad, Patrick J van der Vliet, Karthik T, Nerella Haricharan Goud, William L Macken, Stephanie Efthymiou, Shruthi Nair, Deepak Menon, Ritu Shree, Sireesha Yareeda, Rohit Bhatia, Lindsay A Wilson, Henry Houlden, Michael G Hanna, Enrico Bugiardini, Silvère M van der Maarel, Venugopalan Y Vishnu*

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3.027P The need for and experiences with psychosocial support among adolescents and young adults at the multidisciplinary facioscapulohumeral muscular dystrophy (FSHD) outpatient clinic of radboudumc

Nathaniel Rasing, Ilse Karnebeek, Marnix van Bruggen, Anke Lanser, Nicole B.M. Voet, Karlien Mul, Edith H.C. Cup, ***Nicol Voermans***[1]

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3.028P Long-term trajectories of respiratory function in Facioscapulohumeral Muscular Dystrophy type 1 (FSHD1): a single-center 20-year longitudinal cohort study

Giulio Gadaleta[1], *Liliana Vercelli, Fulvia Ribolla, Guido Urbano, Enrica Rolle, Eleonora Leopizzi, Silvia Boschi, Paolo Solidoro, Tiziana Mongini*

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Poster Session 3: 3.025eP-3.029eP, 3.029P-3.038P: FSHD - Imaging and outcome measures

3.025eP A smartphone-based 30-second sit-to-stand test of lower limb strength in facioscapulohumeral muscular dystrophy

Rachele Rossanigo, Jonathan Street, Francesco Ia Pietra, Alejandro Ena, Daniel Nicoll, Ross Barnes, Óscar Reyes, Shibeshish Belachew, Tecla Bonci, ***Claudia Mazzà***[1,2], *Channa Hewamadduma*

[1]*Indivi AG, Zürich, United Kingdom,* [2]*School of Mechanical, Aerospace and Civil Engineering, University of Sheffield, Sheffield, United Kingdom*

3.026eP Mental and intellectual neuro development in childhood-onset FSHD study: the MINDS-FSHD study protocol

Katy de Valle[1,2,3], *Marloes van Lieshout, Peter Anderson, Corrie Erasmus, Nicol Voermans, Hugh McMillan, Ian Woodcock, Claire Hogan, Chelsea Finchett, Louise Crowe*

[1]*The Royal Children's Hospital Melbourne, Department of Physiotherapy (RCH), Melbourne, Australia,* [2]*The Royal Children's Hospital Melbourne, Department of Neurology, Melbourne, Australia,* [3]*Murdoch Children's Research Institute, Clinical Sciences, Melbourne, Australia*

3.027eP Quantitative imaging outcome measures of muscle contractility in facioscapulohumeral muscular dystrophy using speckle-tracking ultrasound and multiparametric MRI: preliminary results

Odette Van Iersel[1], *Helena TM Boon, Harm Weekenstroo, Jonne Doorduin, Donnie Cameron, Nens van Alfen*

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3.028eP Serum neurofilament light chain mediates the association between blood cadmium exposure and reduced grip strength: evidence from NHANES 2013–2014

Yali Hong[1], *Fuze Zheng, Xinrong Xu, Zhiqiang Wang*

[1]*The First Affiliated Hospital Of Fujian Medical University, Fuzhou, China*

3.029eP Investigating baseline DEXA lean tissue mass as a predictor of longitudinal functional decline in facioscapulohumeral muscular dystrophy

Elizabeth Young[1], *Leo Wang, Maya Hatch, Jason Barber, Doris Leung, Kathryn Wagner, Katy Eichinger, Michaela Walker, Leann Lewis, William Martens, Karlien Mul, Valeria Sansone, Perry Shieh, Bakri Elsheikh, Russell Butterfield, Nicholas Johnson, Carmelo Messina, Elena Carraro, Jeff Statland, Resolve Investigators of the FSHD CTRN*
[1]Department of Neurology, University of Washington, Seattle, United States

3.029P Profiling facioscapulohumeral muscular dystrophy phenotypes by combining muscle MRI and gait analysis: an integrative approach

Eleonora Torchia[1], *Francesco Rocca, Gabriele Denza, Marco Pelliccioni, Sara Bortolani, Valerio Trombetta, Sara De Angelis, Beatrice Ravera, Carmine Di Marco, Davide Marchese, Mauro Monforte, Giorgio Tasca, Giuseppe Vannozzi, Guerrino Rosellini, Enzo Ricci*
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3.030P Self-reported disease burden in childhood-onset FSHD - measurement properties of the pediatric FSHD-health index (FSHD-HI Peds)

Katy de Valle[1,2,3], *William Paterson, William Stronach, Ian Woodcock, Chad Heatwole, Tara Fitzgerald, Nicole Sheers*
[1]The Royal Children's Hospital Department of Physiotherapy (RCH), Melbourne, Australia, [2]Murdoch Children's Research Institute (MCRI), Melbourne, Australia, [3]University of Melbourne Department of Physiotherapy, Melbourne, Australia

3.031P Muscle ultrasound as a potential biomarker in facioscapulohumeral muscular dystrophy: a 10-year follow-up study

Eline Boon[1], *Odette Van Iersel, Karlien Mul, Corrie Erasmus, Nicole Voet, Nicol Voermans, Sanne Vincenten, Nens Van Alfen*
[1]Department of Neurology, Donders Institute for Brain, Cognition and Behaviour, Radboud University Medical Center, Nijmegen, The Netherlands. , Nijmegen, Netherlands

3.032P Assessing functional performance in facioscapulohumeral muscular dystrophy (FSHD) using the glittre activities of daily living test: a pilot study

Ayse Nur Ozdag Acarli[1], *Ecenur Atli, Cagri Ulukan, Ilaha Bakirli, Yusuf Sarican, Havvanur Albayrak, Aysegul Ketenci, Piraye Oflazer*
[1]Koc University, School of Medicine, Department of Neurology, Istanbul, Turkey

3.033P Tracking disease progression in FSHD: change after 1-year of muscle MRI and functional clinical outcomes comparing outcome responsiveness and sensitivity

Mary Foltz[1], *Markus Karlsson, Per Widholm, Jonathan Pini, Angela Puma, Luisa Villa, Michele Cavali, Andra Ezaru, Guillaume Bassez, Benjamin Marty, Teresinha Evangelista, Romain Thomas, Löc Danjoux, Céline Tard, Sabrina Sacconi*
[1]AMRA Medical AB, Linköping, Sweden

3.034P Muscle fat fraction (MFF) progression in facioscapulohumeral muscular dystrophy (FSHD) assessed by whole-body MRI: prognostic factors identified via machine learning

Lucienne Ronco, ***Mary Foltz***[1], *Shuang Wang, Gautam Sajeev, Anyu Zhu, Per Widholm, Andree-Anne Fournier, Amanda Rickard, James Signorovitch*
[1]AMRA Medical AB, Linköping, Sweden

3.035P Progression dynamics and disability milestones in childhood-onset FSHD

Beatrice Ravera[1], *Carmine Di Marco, Ariele Barreto-Haagsma, Sara Bortolani, Eleonora Torchia, Robert Muni-Lofra, Jordi Diaz-Manera, Michela Guglieri, Chiara Marini-Bettolo, Volker Straub, Daniela Leone, Marika Pane, Eugenio Mercuri, Enzo Ricci, Mauro Monforte, Giorgio Tasca*
[1]Università Cattolica Del Sacro Cuore, Rome, Italy

3.036P Motor outcomes to validate evaluations in pediatric facioscapulohumeral muscular dystrophy (MOVE Peds): baseline characteristics for the first twenty participants enrolled

Ian Woodcock[1,2], *Michaela Walker, Brianna Brun, Russell Butterfield, Rebecca Clay, Katy de Valle, Kate Eichinger, Seth Friedman, Maya Hatch, Jianghua He, Natalie Katz, Doris Leung, Leanne Lewis, Katherine Matthews, Rabi Tawil, Jeffrey Statland*
[1]Royal Children's Hospital, Melbourne, Australia, [2]Murdoch Children's Research Institute, Melbourne, Australia

3.037P Electrical impedance myography mirrors MRI and transcriptomic markers of muscle health in facioscapulohumeral muscular dystrophy

Seward Rutkove[1], *Leo Wang, Buket Sonbas-Cobb, Olivia DuCharme, Dennis Shaw, Michaela Walker, Katy Eichinger, Leann Lewis, Rabi Tawil, Johanna Hamel, Karlien Mul, Silvia Blemker, Stephen Tapscott, Seth Friedman, Jeffrey Statland*
[1]Beth Israel Deaconess Medical Center/Harvard Medical School, Boston, MA, United States,

3.038P Quality of life, psychological burden and its correlation with motor function in facioscapulohumeral muscular dystrophy (FSHD): a cross-sectional nationwide study

Andi Nuredini[1], *Giorgia Varallo, Riccardo Cuoghi Costantini, Roberto D'Amico, Sandro Rubichi, Maristella Scorza, Rossella Tupler*
[1]University of Modena and Reggio Emilia, Modena, Italy

Poster Session 3: 3.030eP, 3.039P-3.047P: FSHD - Pathophysiology of NMDs

3.030eP Imaging mass cytometry reveals spatial signatures distinguishing FSHD1 and FSHD2 despite shared clinico-pathological profiles

Bruna Moreira de Souza Proença[1,2], *Sarah Léonard-Louis, Julian Dal Cin, Bérénice Tendrel, Béatrice Labella, Marion Masingue, Anthony Behin, Frédéric Fer, Édouard Berling, Teresinha Evangelista, Yves Allenbach, Tanya Stojkovic, Olivier Benveniste*
[1]*Hospital das Clínicas, Faculty of Medicine, University of São Paulo, São Paulo, Brazil,* [2]*INSERM Centre for Research in Myology UMRS 974, Sorbonne University, Paris, France*

3.039P Resolving diagnostic challenges in facioscapulohumeral muscular dystrophy through integrated genetic and epigenetic profiling

Xinrong Xu[1], *Zhiqiang Wang, Qifang He, Yuhua Lin, Yali Hong*
[1]*Department of Neurology of First Affiliated Hospital, Fujian Medical University, Fuzhou, China,*

3.040P Elucidating the regulatory mechanism that stochastically activates DUX4 in FSHD muscle cells

Mitsuru Sasaki-honda[1], *Hidetoshi Sakurai, Alvaro Rada-Iglesias*
[1]*Ibbtec, University of Cantabria/csic, Santander, Spain*

3.041P Comprehensive methylation profiling facilitates the diagnosis of facioscapulohumeral muscular dystrophy type 2 with novel SMCHD1 variants

Xingyu Xia[1], *Lingchun Liu, Chongbo Zhao, Takako I. Jones, Wenhua Zhu*
[1]*Department of Neurology, Huashan Hospital, Fudan University, Shanghai, China*

3.042P Single-nucleus RNA sequencing defines cellular remodelling in FSHD skeletal muscle

Mohi-uddin Miah[1], *Asmita Saha, Raul Fulea, Philipp Heher, Ariele Barreto Haagsma, Rasya Gokul Nath, Sara Bortolani, Mauro Monforte, Eleonora Torchia, Enzo Ricci, Simon Cockell, Michela Guglieri, Chiara Marini Bettolo, Jordi Diaz Manera, Volker Straub, Giorgio Tasca*
[1]*John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, Newcastle Upon Tyne, United Kingdom*

3.043P Analysing potential novel disease-modifying genes in facioscapulohumeral muscular dystrophy type 2

Mathilde Sanson[1], *Elise N. Engquist, Jiani Qian, Joshua Greig, Peter S. Zammit*
[1]*King's College London, London, United Kingdom*

3.044P Epigenetic dysregulation in FSHD by modeling epigenetic dynamics during early embryonic development

Takumi Kishimoto[1], *Mitsuru Sasaki-Honda, Mio Iwasaki, Yasuhiro Takashima, Hidetoshi Sakurai*
[1]*Center for iPS Cell Research and Application, Kyoto University, Kyoto, Japan*

3.045P Autophagy is perturbed but rescuable in facioscapulohumeral muscular dystrophy

Peter Zammit[1], *Joshua Greig, Philipp Heher*
[1]*King's College London, London, United Kingdom*

3.046P Bridging the diagnostic gap in FSHD with advanced genomic tools

Charlotte Tardy, Jean Philippe Trani, Loeva Morin, Nathalie Eudes, ***Frederique Magdinier***[1]
[1]*Marseille Medical Genetics, Marseille, France*

3.047P Integrated clinico-histological, transcriptomic, and spatial proteomic characterization of FSHD1 and FSHD2: a monocentric cohort study

Bruna Moreira de Souza Proença[1,2], *Sarah Léonard-Louis, Julian Dal Cin, Bérénice Tendrel, Béatrice Labella, Marion Masingue, Anthony Behin, Frédéric Fer, Édouard Berling, Teresinha Evangelista, Yves Allenbach, Tanya Stojkovic, Olivier Benveniste*
[1]*Hospital das Clínicas, Faculty of Medicine, University of São Paulo, São Paulo, Brazil,* [2]*INSERM Centre for Research in Myology UMRS 974, Sorbonne University, Paris, France*

Poster Session 3: 3.031eP, 3.048P-3.049P: FSHD – Treatments

3.031eP Rationale and design of a safety, tolerability, and long-term efficacy study of SAT-3247 in facioscapulohumeral muscular dystrophy (FSHD)

Ryan Mitchell, Phil Lambert, Michael Rudnicki, ***Wildon Farwell***[1], *Kristiana Salmon*
[1]*Satellos Bioscience, Toronto, Canada*

3.048P A chemically novel antisense oligonucleotide DUX4 inhibitor for FSHD: pharmacokinetics/biodistribution in rats, monkeys and mouse FSHD disease model

Eva Chin[1], *Lars Nordholm, Chie Fukunaga, Yuko Ogasawara, Norihiko Iwazaki, Tetsuhiro Kakimoto, Hajime Aihara, Christoph Rosenbohm, Yann Tessier, Kathryn R. Wagner, Klaus Jensen*
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3.049P Metformin enhances skeletal muscle differentiation and function in facioscapulohumeral muscular dystrophy models

Joshua Greig[1], *Philipp Heher, Peter Zammit*
[1]*King's College London, London, United Kingdom*

Poster Session 3: 3.032eP-3.036eP, 3.050P-3.057P: Glycogenoses

3.032eP Expanding the molecular basis of glycogen storage disease due to muscle phosphorylase kinase deficiency: a novel candidate gene through exome sequencing

Giovanni Vicidomini[1], *Pasquale Di Letto, Margherita Scarpato, Francesca Pia Palmieri, Valerio Bonolis, Gaia Esposito, Esther Picillo, Marianna Scutifero, Lucia Ruggiero, Dario Zoppi, Filippo Maria Santorelli, Stefano Doccini, Giulio Piluso, Annalaura Torella, Vincenzo Nigro*
[1]*Università degli Studi della Campania "Luigi Vanvitelli", Naples, Italy*

3.033eP Delivery of acid α-glucosidase by muscle-targeting extracellular vesicles: a new road for Pompe disease treatment

Mafalda Cacciottolo[1], *Li-En Hsieh, Daniel Leychkis, Wenny Wong, Bailey Murphy, Huilan Lin, Christy Lau, Nicole Bee, Linda Marban, Kristi Elliott, Minghao Sun*
[1]*Capricor Therapeutics, San Diego, United States*

3.034eP Preparing the first in-human study of lentiviral gene therapy for Pompe disease

Pim Pijnappel[1], *Giacomo Zundo, Fabio Catalano, Tessa Huizer, Wassim Ouhammou Taouni, Hannerieke van den Hout, Nadine van der Beek, Sofieke de Wilde, Michael Rothe, Axel Schambach, Isabel Gordaliza, Karin Pike-Overzet, Arjan Lankester, Frank Staal, Dirk van Asseldonk*
[1]*Erasmus MC University Medical Center, Rotterdam, Netherlands*

3.035eP ATB200-03/07 Danish subpopulation: 4-year efficacy and safety of cipaglucosidase alfa plus miglustat in adults with late-onset Pompe disease

Henning Andersen[1], *Ali Hariri, Emma Flordal Thelander, Sofia Nordin, Vipul Jain*
[1]*Department of Neurology, Aarhus University Hospital, Aarhus N, Denmark*

3.036eP Applying an integrated multiparametric digital platform to Pompe disease

Francesca Torri[1], *Francesco Sansone, Alessandro Tonacci, Raffaele Conte, Giulia Ricci, Gabriele Siciliano*
[1]*Department of Clinical and Experimental Medicine, University of Pisa, Pisa, Italy*

3.050P Real-world implementation, safety and outcomes of new enzyme replacement therapies in Pompe disease: data from the Spanish registry

Sonia Segovia Simon[1,2], *Cristina Puig, Cristina Domínguez-González, Nuria Muelas, Carmen Paradas, Rafael Genaro, Maria Teresa Gomez, Juan Restrepo, María Camprodon, Velina Nedkova, Germán Morís, Jorge Garcia Garcia, David Reyes, Maria Dolores Mendoza, Jorge Alonso-Pérez, Jeidy Marcela Henao, Jordi Diaz Manera*
[1]*John Walton Muscular Dystrophy Research Center, Newcastle, United Kingdom,* [2]*Newcastle University, Newcastle Upon Tyne, United Kingdom*

3.051P Design of a phase 2 trial of S-606001 as add-on substrate reduction therapy (SRT) to enzyme replacement therapy (ERT) in late onset Pompe disease

Yasuhiro Tabata, Prasant Chandran, Nirvana Raghubir, Polina Elistratova, Christopher Chatham, Aya Kuwata, Shinji Kakudo, ***Simon Portsmouth***[1]
[1]*Shionogi Inc., Florham Park, United States*

3.052P Developing a 3D in vitro model of skeletal muscle involvement in late-onset Pompe disease for CRISPR-based therapy testing

Francesca Torri[1], *Federica Zinghirino, Arta Aghaeipour, Valentina Lionello, Alessia Cavazza, Francesco Saverio Tedesco, Sara Benedetti*
[1]*Infection, Immunity and Inflammation Dept, UCL GOS Institute of Child Health Zayed Centre for Research into Rare Disease in Children, London WC1N 1DZ, UK, London, United Kingdom*

3.053P Spatial transcriptomics reveals early and progressive molecular changes in late-onset Pompe disease muscle

Alexandra Monceau[1], *Eduard Gallardo, Montse Olivé, Aurelio Hernández-Lain, Carmen Paradas, Eloy Rivas, Cristina Domínguez-González, George Papadimas, Constantinos Papadopoulos, Margarita Chrysanthou-Piterou, Anna Kostera-Pruszczyk, Biruta Kierdaszuk, James Lilleker, Mark Roberts, Olimpia Musumeci, James Clark, Esther Fernández-Simon, Antonio Toscano, Jordi Díaz-Manera*
[1]*Newcastle University, Newcastle, United Kingdom*

3.054P Advanced modelling of infantile-onset Pompe disease to develop next-generation genetic therapies

Arta Aghaeipour[1,2], *Francesca Torri, Pauline Vidal, Fanny Collaud, Shalini Reddy, Manju Kurian, Giuseppe Ronzitti, Francesco Saverio Tedesco, Sara Benedetti*

[1]*Infection, Immunity and Inflammation Research and Teaching Department, UCL GOS Institute of Child Health, London, United Kingdom,* [2]*The Francis Crick Institute, London, United Kingdom*

3.055P Baby-COMET study: Safety and efficacy of avalglucosidase alfa in treatment-naïve participants with infantile-onset Pompe disease

Yin-Hsiu Chien[1], *Andreas Hahn, Lijun Fu, Robert J. Hopkin, Sihoun Hahn, Peter Witters, Katherine Denlinger, Kristina An Haack, Olivier Huynh-Ba, Carlota Garcia, Kelly George, Tianyue Zhou, Madhurima Uppara Kowthalam, Priya Kishnani*

[1]*Department of Medical Genetics, National Taiwan University Hospital, Taipei, Taiwan*

3.056P Long-term (4-year) efficacy and safety of cipaglucosidase alfa plus miglustat in adults with late-onset Pompe disease treated from PROPEL baseline

Mark Roberts[1], *Drago Bratkovic, Kristl G Claeys, Jordi Díaz-Manera, Tahseen Mozaffar, Michael I Polkey, Nadine AME van der Beek, Benedikt Schoser, Antonio Toscano, Fred Holdbrook, Vipul Jain, Pilar Moore, Kinesha O'Brien-Prince, Priya S Kishnani*

[1]*Salford Royal NHS Foundation Trust, Salford, United Kingdom*

3.057P Baseline disease burden and real-world monitoring practices: global insights from the followME Pompe Journey registry

Lindsay Alfano[1], *Henning Andersen, Kristl Claeys, Brad Crittenden, Stephanie DeArmey, Patrick Deegan, Robin Lachmann, Paul McIntosh, Eugen Mengel, Mark Roberts, Thomas Scherer, Fred Holdbrook, Vera Gielen, Vipul Jain, William Kerr, Kinesha O'Brien-Prince, Sophie Clarke, Stephan Wenninger*

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Poster Session 3: 3.037eP-3.066eP, 3.058P-3.098P: Idiopathic inflammatory myopathies

3.037eP Development of the IMNM functional rating scale: a disease-specific clinical outcome assessment for immune-mediated necrotizing myopathy

Siddhant Pratap[1], *Tahseen Mozaffar*

[1]*University of California, Irvine, Irvine, CA, United States*

3.038eP Clinical characteristics, healthcare utilization, and unmet needs of patients with inclusion body myositis: a nationwide questionnaire survey

Satoshi Yamashita[1], *Tomoyuki Otsu, Hiroyuki Murai*

[1]*International University of Health and Welfare Narita Hospital, Narita, Japan*

3.039eP A case series of paraneoplastic anti-TIF1-γ antibody-positive dermatomyositis: experience from an Indonesian tertiary healthcare center

Carmelia Anggraini[1,2], *Luh Ari Indrawati, Shierly Novitawati Sitanaya, Adrian Ridski Harsono, Dinda Larastika Riyanto, Nurul Fadli, Winnugroho Wiratman, Ahmad Yanuar Safri, Astri Budikayanti, Fitri Octaviana, Manfaluthy Hakim*

[1]*Departement of Neurology, Faculty of Medicine, Universitas Indonesia, Jakarta, Indonesia,* [2]*Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia*

3.040eP No association between inclusion body myositis and cancer risk in a population-based case-control study

Pierre Larsson[1,2], *Ulrika Lindgren*

[1]*University of Gothenburg, Gothenburg, Sweden,* [2]*Sahlgrenska University Hospital, Gothenburg, Sweden*

3.041eP Delayed diagnosis, high disease burden, and therapeutic outcome in anti-signal recognition particle immune-mediated necrotizing myopathy (anti-SRP IMNM): findings from an Indonesian registry

Faradillah Mutiani[1,2], *Shierly Novitawati Sitanaya, Dinda Larastika Riyanto, Adrian Ridski Harsono, Nurul Fadli, Winnugroho Wiratman, Ahmad Yanuar Safri, Astri Budikayanti, Fitri Octaviana, Manfaluthy Hakim, Luh Ari Indrawati*

[1]*Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia,* [2]*Faculty of Medicine Universitas Indonesia, Jakarta, Indonesia*

3.042eP Clinical characteristics of patients with systemic sclerosis complicated with muscular symptoms: from a single center retrospective cohort data

Shu Sugimoto[1], *Akinori Uruha, Dai Kishida, Yoshiki Sekijima*

[1]*Shinshu University School of Medicine, Matsumoto, Japan*

3.043eP Divergent functional trajectories in inclusion body myositis: sirolimus, methotrexate, and untreated patients across two countries

Bruna Moreira de Souza Proença[1], *André Macedo Serafim da Silva, Edmar Zanoteli, Olivier Benveniste*

[1]*Department of Neurology, Faculdade de Medicina, Universidade de São Paulo (FMUSP), São Paulo, Brazil*

3.044eP Spatial transcriptomics in immune-mediated necrotizing myopathy reveals a specific necrotic signature related to immune-driven fibrosis

Julian Sanchez-Dal Cin[1], *Alexandrine Mahoudeau, Mona Bensalah, Laura Muraine, Bérénice Tendrel, Céline Anquetil, Damien Amelin, Loïs Bolko, Linda Chenane, Franck Letourneur, Angéline Madelaine, Sarah Léonard-Louis, Elisa Negroni, Capucine Trollet, Olivier Benveniste, Yves Allenbach*

[1]*Sorbonne University, INSERM, APHP, Paris, France*

3.045eP Unmet needs within current outcome measures for immune-mediated necrotizing myopathy

Siddhant Pratap[1], *Tahseen Mozaffar*

[1]*University of California, Irvine, Irvine, CA, United States*

3.046eP Case series of granulomatous myositis: a descriptive study of a rare inflammatory myopathy

Iñigo Rojas-marcos Rodriguez Quesada[1], *Ignacio Lopera-Rodriguez, Francisco-Javier Gómez-Fernández, Eloy Rivas-Infante, Carmen Paradas*

[1]*Hosp General Virgen Del Rocio : Serv Neurologia, Sevilla, Spain*

3.047eP Clinical, radiological, and pathological characteristics of pediatric anti-HMGCR myopathy in Korea

Jee Min Kim[1], *Seungbok Lee, Soo Yeon Kim, Yun Jeong Lee, Eun Na Kim, Anna Cho, Byung Chan Lim, Jong-Hee Chae*

[1]*Department of Genomic Medicine, Seoul National University Hospital, Seoul, South Korea,*

3.048eP Relapse characteristics in seropositive immune-mediated necrotizing myopathy: steroid tapering, creatine kinase, and immunosuppressive therapy in a retrospective cohort

Tri Si Le[1], *Vinh Khang Nguyen*

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3.049eP Muscle involvement in connective tissue disease

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3.050eP The diagnostic utility of p62 immunohistochemistry in immune-mediated necrotizing myopathy

Kotaro Matsumoto[1,2,3], *Yumi Yamamoto, Yusuke Baba, Rui Shimazaki, Shinichiro Hayashi, Satoru Noguchi, Kazuma Sugie, Ichizo Nishino*

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3.051eP p62 aggregation in anti-mitochondrial M2 antibody-positive myositis: a single-center retrospective case series

Yukako Nishimori[1], *Nobuyuki Eura, Kazuma Sugie*

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3.053eP Fantastic myositis and how to find it: a clinico-serological profile of idiopathic inflammatory myopathy from a national referral hospital in Indonesia

Shierly Novitawati Sitanaya[1,2], *Ahmad Yanuar Safri, Dinda Larastika Riyanto, Nurul Fadli, Winnugroho Wiratman, Adrian Ridski Harsono, Astri Budikayanti, Manfaluthy Hakim, Fitri Octaviana, Luh Ari Indrawati*

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3.054eP Clinical characteristics and management of anti-HMGCR immune-mediated necrotizing myopathy: a case series from Indonesia

Manfaluthy Hakim[1,2], *Dinda Larastika Riyanto, Adrian Ridski Harsono, Nurul Fadli, Winnugroho Wiratman, Ahmad Yanuar, Astri Budikayanti, Fitri Octaviana, Luh Ari Indrawati*

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3.055eP Immunofluorescence-based identification of muscle-reactive antibodies in sporadic late-onset nemaline myopathy (SLONM)

Francesca Forcina[1], *Antonio Lauletta, Gioia Merlonghi, Matteo Garibaldi, Eduard Gallardo Vigo*

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3.056eP Sporadic late-onset nemaline myopathy with monoclonal gammopathy presents a long-term response to a bortezomib-lenalidomide regime but not to autologous stem cell transplantation

Iñigo Rojas-marcos Rodriguez Quesada[1,2], *Eusebio Martín-Chacón, Ignacio Lopera, Javier Gómez-Fernández, Eloy Rivas, Macarena Cabrera, Carmen Paradas*

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3.057eP Journey Through the NXP2 dermatomyositis spectrum: insights gained from a case
Mustafa Cam[1], *Yiğit Can Güldiken, Abdulsamed Kılıç*
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3.058eP Dermatomyositis in the context of small cell lung cancer: diagnostic challenges and therapeutic insights
Yigit Can Guldiken[1], *Batool Achmar*
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3.059eP Intramuscular heterotopic ossification: an unusual presentation of juvenile-onset anti-NXP2-related myopathy
Aldrin Martins[1], *Gustavo Freitas Alves de Arruda, Mateus Augusto Rangel Nunes, Ana Mondadori dos Santo, Alisson Pugliesi, Ana Paula Toledo del Rio, Alberto Rolim Martinez, Felipe Franco, Anamarli Nucci, Marcondes Cavalcante França Jr*
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3.060eP Acute unilateral myonecrosis with anti–SRP seropositivity: a case of clinicopathological Discordance and Spontaneous Recovery
Jamie Kok[1], *Joanne Tan*
[1]*National University Hospital, Singapore, Singapore*

3.061eP Paraneoplastic dermatomyositis secondary to ovarian DLBCL: clinical presentation and diagnostic challenges
Anna Lewandowska-Polak[1], *Milena Kaszubska, Szymon Lis, Olga Brzezinska, Magdalena Opinc, Aleksandra Opinc-Rosiak, Joanna S. Makowska*
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3.062eP Infantile-onset anti-signal recognition particle antibody-positive immune-mediated necrotizing myopathy: a case report
Ayaka Ohno[1], *Takashi Kurashige, Yoshiyuki Kobayashi, Takehiko Doi, Kasumi Sasaki, Yuta Eguchi, Yuichi Tateishi, Ichizo Nishino, Satoshi Okada*
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3.063eP Undiagnosed case of progressive myopathy with early respiratory involvement and a candidate variant in the FHL1 gene
Lisanna Põlluaas[1,2], *Piret Mertsina, Liis Väli, Sanna Puusepp, Siiri Sarv, Katrin Õunap*
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3.064eP Orbital myositis in hyperthyroidism
Nadhila Nur Shafitha[1], *Christopher Silman, Luh Ari Indrawati*
[1]*Department of Neurology, Mandaya Royal Hospital Puri, Tangerang, Indonesia*

3.065eP Anti-NXP2 dermatomyositis with fatal gastrointestinal bleeding: an autopsy case report
Koichiro Watanabe[1], *Hajime Iwata, Hiroshi Saito, Nozomu Abe, Akihito Hao, Satoko Uruha, Eisuke Takamasu, Haruka Okada, Keisuke Ishizawa, Akinori Uruha*
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3.066eP Clinical features of adult late-onset nemaline myopathy associated with connective tissue diseases
Na Ji, Xisoke Wu, Xiaoping Huang, Tingjun Dai, Qi Fang, Chuanzhu Yan, ***Meirong Liu***[1]
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3.058P Clinical utility of comprehensive autoantibody profiling in IIM: a context-based approach to ANA interpretation
Soohyun Kim[1], *Hyung Jun Park, Bum Chun Suh*
[1]*Department Of Neurology, Gangnam Severance Hospital, Yonsei University College Of Medicine, Seoul, South Korea*

3.059P Amyloid myopathy: clinicopathological and morphometrical correlations
Sarah Souvannanorath, ***Baptiste Periou***[1,2], *Mary Marcela Araujo, Karolina Hankiewicz, Catherine Heyraut-Blanchet, Violaine Plante-Bordeneuve, Claire Lefeuvre, Thibaut Damy, Amira Zaroui, Gianmarco Severa, Léonard Féasson, Edoardo Malfatti, François-Jérome Authier*
[1]*Paris Est Créteil University, Créteil, France,* [2]*Henri Mondor University Hospital, Créteil, France,*

3.060P Current landscape of immunotherapy access for idiopathic inflammatory myopathy in Indonesia: a national survey study

Muhammad Tito Anugerah Ramadhan[1,2], *Faradillah Mutiani, Muhammad Faruq Abdan Syakuron, Ahmad Yanuar Safri, Dinda Larastika Riyanto, Adrian Ridski Harsono, Nurul Fadli, Winnugroho Wiratman, Astri Budikayanti, Fitri Octaviana, Manfaluthy Hakim, Luh Ari Indrawati*

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3.061P Clinical outcome trajectories in inclusion body myositis stratified by anti-NT5c1A serostatus

Karina Bjazevic[1], *Isela Hernandez, Namita Goyal, Spencer Matthews, Olimpia Carbunar, Miriam Freimer, Mazen Dimachkie, Colin Quinn, Thomas Lloyd, Payam Mohassel, Conrad Weihl, Aziz Shaibani, Leo Wang, Nizar Chahin, Anthony Amato, Matthew Wicklund, Stacy Dixon, Perry Shieh, Tahseen Mozaffar, INSPIRE-IBM Study Group*

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3.062P Plasma proteomic profiling reveals distinct molecular signatures in Idiopathic inflammatory myopathies: a multiplex proximity extension assay study

Yunjung Choi[1], *Soo-Hyun Kim, Young-Chul Choi, Hyung Jun Park*

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3.063P Prevalence of osteoporosis and pathological fractures in anti-mitochondrial antibody–positive myositis: results of a nationwide survey

Meiko Maeda[1], *Takuya Kawahara, Akatsuki Kubota, Jun Shimizu, Tatsushi Toda*

[1]*Department of Neurology, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan,*

3.064P Hidden malnutrition predicts mortality in idiopathic inflammatory myopathies despite prevalent overweight and obesity

Joanna Makowska[1], *Aleksandra Opinc-Rosiak, Ewelina Kulesza, Kacper Pawlak, Agnieszka Mikosińska, Olga Brzezińska, Bartosz Stępień, Anna Lewandowska-Polak*

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3.065P Diagnostic performance of established criteria for inclusion body myositis: a preliminary clinicopathologic study

Nur Atikah Tajul Lile[1], *Louisa Kent, Monika Hofer, Stefen Brady*

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3.066P Clinicopathological profile of eosinophilic fasciitis: a retrospective cohort study from a neuromuscular disorder center in China

Bing Zhao[1], *Xiaoyuan Wang, Kai Shao, Chuanzhu Yan*

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3.067P A retrospective study on the efficacy of intravenous immunoglobulin therapy in inclusion body myositis

Megumi Kubota[1], *Hotake Takizawa, Yasushi Oya, Yuji Takahashi*

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3.068P Long-term follow-up and risk factors for relapse in patients with anti-HMGCR necrotizing myopathy

Orane Demuynck, ***Olivier Benveniste***[1]

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3.069P Clinicopathological characteristics of polymyositis with mitochondrial pathology (PM-Mito)

Shogo Komaki[1], *Akatsuki Kubota, Meiko Maeda, Jun Shimizu, Wataru Satake*

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3.070P Clinical, imaging, and muscle pathological characteristics of different subtypes of juvenile idiopathic inflammatory myopathies

Cuijie Wei[1], *Gen Ge, Xingzhi Chang, Hui Xiong*

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3.071P Broad pathological and radiological heterogeneity in anti-U1-RNP antibody-positive myositis

Shuichiro Amano[1,2,3], *Sota Shiomi, Ai Yamanaka, Shinichiro Hayashi, Satoru Noguchi, Kazuma Sugie, Ichizo Nishino*

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3.072P Immunophenotyping of patients with inclusion body myositis reveals immune cell imbalance: INSPIRE-IBM study

Philip Farahat[1], *Connor Philips, Isela Hernandez, Marie Wencel, Namita Goyal, Tahseen Mozaffar*
[1]*University Of California, Irvine, Irvine, United States*

3.073P Development and validation of the MDA-FI score: a simple prognostic tool at diagnosis in anti–MDA5 dermatomyositis

Quentin Astouati, Valentine Pagis, Lucas Pacoureau, Yann Nguyen, Pierre Bay, Antoine Roux, Hubert De Boysson, Laure Gallay, Vincent Cottin, Benjamin Terrier, Alain Meyer, Charles Cerf, Mathilde Neuville, Baptiste Hervier, Hilario Nunes, Eric Hachulla, David Launay, Yurdagül Uzunhan, Olivier Benveniste, ***Yves Allenbach***[1]
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3.074P Long-term safety and efficacy of efgartigimod in idiopathic inflammatory myopathy: phase 2 ALKIVIA and open-label extension ALKIVIA+ trial results

Angela Rosenbohm[1], *Rohit Aggarwal, Anthony A. Amato, Ingrid E. Lundberg, Andrew L. Mammen, Jiří Vencovský, Despoina Papadopoulou, Paul Duncombe, Bas van der Woning, Hector Chinoy*
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3.075P MDA5-seropositive dermatomyositis in Dr Cipto Mangunkusumo hospital Indonesia

Luh Indrawati[1,2], *Dinda Riyanto, Adrian Harsono, Nurul Fadli, Winnugroho Wiratman, Ahmad Yanuar, Astri Budikayanti, Fitri Octaviana, Manfaluthy Hakim*
[1]*Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia,* [2]*Faculty of Medicine Universitas Indonesia, Jakarta, Indonesia*

3.076P Clinical characteristics and cancer risk of anti-OJ antisynthetase syndrome: a cohort comparative study and a systematic literature review

Yves Allenbach[1], *Valentin Pillot, Segolene Tocquet, Jean Luc Charuel, Sarah Tansley, Samia Boussouar, Pierre Yves Brillet, Hui Lu, Dylan Vellas, Sami Tayb Boulahfa, Sarah Leonard Louis, Yurdagul Uzunhan, Pascale Ghillani-Dalbin, Makoto Miyara, Loïc Baudelet, Maxime Ilzkovitz, Carole Nagant, Anne Marie Bouvier, Valerie Jooste, Olivier Benveniste*
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3.077P Galectin-3-mediated macrophage and FAP interactions that regulate fatty fibrosis in IBM muscle

Diego Jaime[1], *Tahseen Mozaffar, Namita Goyal, Apoorva Nagendra, Isela Hernandez, Wei Zhao, Qing Nie, Armando Villalta*
[1]*University of California, Irvine, Irvine*

3.078P Clinico-pathological and serological correlation of immune mediated necrotizing myopathy patients in Indian cohort

Mehar Sharma[1], *Bandana Jassal, Vaishali Suri, Venugopalan Y Vishnu, Danveer Bhadu*
[1]*Agilus Lab FMRI Gurugram, Delhi, India*

3.079P Characterising muscle-infiltrating B cells in inclusion body myositis

Nataliya Slater[1,2,3], *Yueran Li, Abha Chopra, Jerome Coudert, Merrilee Needham*
[1]*Murdoch University, Perth, Australia,* [2]*Perron Institute for Neurological and Translational Research, Perth, Australia,* [3]*Notre Dame University, Perth, Australia*

3.080P The role of extracellular vesicles containing TDP-43 and DJ-1 in the pathogenesis of inclusion body myositis

Benedict Voss[1,2], *Vera Dobelmann, Lyn Eigenfeldt, Andreas Hentschel, Basant Thakur, Venkatesh Chetty, Lorenzo Maggi, Alessandra Ruggieri, Giorgia Riolo, Sara Gibertini, Andreas Roos, Tobias Ruck*
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3.081P Intercellular adhesion molecule 1 (ICAM-1) in muscle and blood is a promising novel biomarker to diagnose and monitor myositis

Leonie Kather[1], *Alexander Mensch, Melanie Huber, Angela Roth, Carina Schreiyäck, Vincent Umathum, Heidrun Krämer-Best, Ulf Müller-Ladner, Marek Bartkuhn, Markus Otto, Elena Neumann, Anne Schänzer*
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3.082P Proteomic characterization of COX-SDH abnormal fibres in polymyositis with mitochondrial pathology

Karsten Krause[1,2], *Christopher Nelke, Tobias Ruck, Andreas Hentschel, Andreas Roos, Felix Kleefeld*
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3.083P Interferon subtypes direct autophagy: toward distinct therapeutic pathways in idiopathic immune myopathies

Ludovic Martin[1], *Matthieu Doudard, Lola Albertella, Ana Šarić, Olivier Conscience, Baptiste Periou, Sarah Souvannanorath, Edoardo Malfatti, François Jérôme Authier*

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3.084P Defining a unified SSc myopathy spectrum: quantitative histopathology may help predict clinical severity

Louai ZAIDAN, Noémie Le Gouellec, Baptiste Periou, Fred Relaix, Luc Mouthon, Eric Hachulla, Nadia Oubaya,

Francois Jerome Authier[1]

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3.085P Cardio-independent troponin T elevation in myopathies and myositis

Angela Rosenbohm[1], *Sabrina Ege, Dominik Buckert, Nam Nguyen-Younossi, Jochen Weishaupt*

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3.086P Shared molecular signatures of granulomas across sarcoid and non-sarcoid granulomatous myopathies

Guang Ji[1], *Shunsuke Funaguma, Aritoshi Iida, Satoru Noguchi, Shinichiro Hayashi, Ichizo Nishino*

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3.087P The effects of serum, containing myositis-specific autoantibodies, on muscle fiber contractility in immune-mediated necrotizing myopathy

Tom Joseph Kerkhoff[1,2,3], *Sanna Luijcx, Buram Ardiç, Patrycja Stachurska, Haya Abboud, Sanne Evers, Anneke J. Van der Kooi, Eleonora Aronica, Theo Rispens, Joost Raaphorst, Coen Ottenheijm*

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3.088P Effectiveness and safety of rituximab in the treatment of idiopathic inflammatory myopathies at the neurological institute of Thailand

Tanapon Siritananukulwong[1], *Arada. Rojana-udomsart*

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3.089P Comparative evaluation of respiratory assessments in inclusion body myositis from INSPIRE-IBM study

Namita Goyal, Matthews Spencer, Olimpia Carbunar, Miriam Freimer, Mazen Dimachkie, Colin Quinn, Thomas E. Lloyd, Payam Mohassel, Conrad Weihl, Aziz Shaibani, Leo Wang, Nizar Chahin, Anthony A. Amato, Matthew Wicklund, Stacy Dixon, Perry Shieh, Laura Herbelin, Richard E. Barohn, Tahseen Mozaffar, ***Marie Wencel***[1]

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3.090P Characterization of interferon signature in antisynthetase syndrome associated myositis

Dilbe Dari[1], *Laure Gallay, Iago Pinal-Fernandez, Maria Casal-Dominguez, Andrew Mammen, Olivier Benveniste, Hans-Hilmar Goebel, Natalie Streichenberger, Andreas Roos, Tobias Ruck, Werner Stenzel, Corinna Preusse*

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3.091P A novel eotaxin/CCR3–mediated mechanism linking macrophage infiltration with viral entry and muscle degeneration in inclusion body myositis

Giorgia Riolo[1,2], *Alex Vicino, Marta Cheli, Franco Salerno, Sara Gibertini, Alessandra Ruggieri, Giada Maria Giunta, Eliana Iannibelli, Alessandra Carnazzi, Lucia Nicolini De Gaetano, Massimo Costanza, Silvia Bonanno, Stefania Marcuzzo, Paola Cavalcante, Lorenzo Maggi*

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3.092P From PM-mito to IBM: longitudinal analysis identifies an early interferon-gamma–driven disease phase

Felix Kleefeld[1], *Alexander Mensch, Karsten Krause, Werner Stenzel, Paula Quint, Corinna Preusse, Menekse Öztürk, Tobias Ruck, Christopher Nelke*

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3.093P A transfer-locomotor axis as a sensitive marker of early functional decline in inclusion body myositis: a longitudinal observational study

Bruna Moreira de Souza Proença[1], *Renata Ferrari Silva, Tatiane Ribeiro Fernandes, Ana Paula dos Anjos Lança, Gabriel Keller, Cristiane de Araújo Martins Moreno, Andre Macedo Serafim da Silva, Edmar Zanoteli*

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3.094P Reclaim-PM: a retrospective reclassification of muscle biopsies diagnosed as polymyositis using contemporary clinico-sero-pathological criteria

Julian Sehne[1,2], *Karsten Krause, Tobias Ruck, Christopher Nelke, Werner Stenzel, Felix Kleefeld*
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3.095P Subtype-specific differences in lipid accumulation and metabolic pathways in immune-mediated necrotizing myopathy

Nathalie Focks[1], *Oliver Baum, Philipp Eisenlohr, Carsten Dittmayer, Corinna Preusse, Werner Stenzel, Felix Kleefeld*
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3.096P Mortality and causes of death in idiopathic inflammatory myopathies: survival study from a nationwide French cohort of 2727 patients

Dylan Vellas[1,2], *Cyril Gitiaux, Ludivine Kouton, Olivier Benveniste, Anne-Sophie Jannot, Yves Allenbach*
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3.097P Juvenile dermatomyositis across diagnostic eras: treatment evolution and long-term outcomes in a 25-year cohort

Mehmet Orhan Erkan, Hülya Ercan Emreol, Mehmet Besrek, Yagmur Bayindir, ***Sibel Öz Yıldız***[1], *Veysel Çam, Dilara Ünal, Özlem Necipoğlu Banak, Hazel Delal Dara Kar, Şule Özkan, Beril Talim, Adalet Elçin Yıldız, Özge Başaran, Yelda Bilginer, Göknur Haliloğlu, Seza Özen, Erdal Sağ*
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3.098P Atypical skin lesions revealing positive anti-HMGCR immune-mediated necrotizing myopathy (IMNM) with severe fasciitis. A case report

Emna Farhat[1,2], *Hicham Tarzi, Souhail Marrak*
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Poster Session 3: 3.067eP-3.068eP: Lipid storage myopathies

3.067eP Growth differentiation factor 15: a valuable biomarker for the diagnosis and prognosis of late-onset form of multiple Acyl-CoA dehydrogenation deficiency

Yuan Sun[1], *Shuyao Tang, Kunqian Ji, Chuanzhu Yan*
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3.068eP Diagnosis by sequencing: PNPLA2 variants masked by genomic complexity guide clinical investigation and management

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[2]*Department of Diagnostic Genomics, PathWest Laboratory Medicine, Perth, Australia*

Poster Session 3: 3.069eP-3.075eP, 3.099P-3.106P: Mitochondrial disorders

3.069eP TANGO2 disease: clinical features, management, and monitoring experience

Laura Carrera-García, Silvia Meavilla, Natalia Julia, Agustín Lujan, Daniel Natera-de Benito, Jesica Expósito Escudero, Roser Colomer, Andrea Greco, Berta Estévez-Arias, Estefanía Martínez, Angels García-Cazorla, Carlos Ortez, Georgia Sarquella-Brugada, ***Andres Eduardo Nascimento Osorio***[1]
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3.070eP Mitochondrial trifunctional protein deficiency may mimic recurrent Guillain-Barré syndrome with episodic weakness

Nagihan Şener, Gizem Ürel Demir, Selman Kesici, Deniz Çağdaş Ayvaz, Yılmaz Yıldız, Dilek Yalnızoğlu, Gökçem Yıldız Sarıkaya, Çağrı Temuçin, ***Göknur Haliloğlu***[1]
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3.071eP Clinical spectrum of mitochondrial complex IV deficiency associated with SCO2 gene mutations: 3 case reports and literature review

Chaoping Hu[1,2], *Yiyun Shi, Lei Zhao, Xihua Li, Lifei Yu, Taosheng Huang*
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3.072eP Cerebellar atrophy as a diagnostic clue in early-onset myopathy: MSTO1-related disease in two siblings
*Hatice Bektas, Ahmet Cevdet Ceylan, **Didem Ardıçlı**[1,2]*
[1]Pediatric Neurology Clinic, Ankara Bilkent City Hospital, Ankara, Turkey, [2]Department of Pediatrics, Division of Pediatric Neurology, Ankara Yıldırım Beyazıt University Faculty of Medicine, Ankara, Turkey

3.073eP Progressive ptosis and ophthalmoplegia with normal lactate revealing mitochondrial myopathy
***Veronica Ivonne Lopez Diaz**[1], Maria Elena Meza-Cano*
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3.074eP Biallelic MCUR1 nonsense mutation associated with vacuolar myopathy and altered mitochondrial calcium signalling
***Anna Maria Haschke**[1], Anja von Renesse, Eugenio Graceffo, Susanne Morales-Gonzalez, Alessandro Prigione, Christoph Hübner, Werner Stenzel, Markus Schuelke*
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3.075eP Megaconial myopathy in a 19-year-old woman with cystic fibrosis
***Beata Sikorska**[1,2], Olena Zimba*
[1]Medical University of Lodz, Lodz, Poland, [2]University Hospital of Krakow, Krakow, Poland

3.099P Proteomic insights into a homozygous SLC25A51 variant in two siblings with suspected mitochondrial disease
***Bülent Kara**[1], Beyzanur Çobanoğlu, Ayten Yazıcı, Özlem Ünal, Naci Çine, Murat Kasap, Gürler Akpınar*
[1]Kocaeli University Medical Faculty Department of Pediatrics, Kocaeli, Turkey

3.100P Identification of cell-type specific adaptations to mitochondrial stress in patient iPSC-derived models of MTRFR mutations
***Kaela O'connor**[1,2], Mariana Zarate-Mendez, Sally Spendiff, Rita Horvath, Hanns Lochmüller*
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3.101P Clinical and genomic spectrum of mitochondrial aminoacyl-tRNA synthetase disorders: a case series from Türkiye
***Ozlem Yayici Koken**[1], Büşra Nur Çavdarlı, Elif Perihan Öncel, Gökçen Öz Tunçer, Ahmet Cevdet Ceylan, Arzu Eroğlu, Nihal Olgaç Dündar, İlknur Erol, Ayşe Aksoy, Esra Manguoğlu, Haluk Topaloğlu*
[1]Akdeniz University, Faculty of Medicine, Department of Child Neurology, Antalya, Turkey

3.102P Large Maf transcription factor family as a therapeutic target for fast myofiber atrophy induced by mitochondrial abnormalities
***Tatsuya Matsumoto**[1], Shunya Sadaki, Tomoki Sato, Shinji Miura, Tomohiko Suzuki, Satoru Takahashi, Ryo Fujita*
[1]University of Tsukuba, Ibaraki, Japan

3.104P In vivo amino acid treatment attenuates phenotype rescue in ears2-/- zebrafish embryos as a treatment for mitochondrial translation deficiencies
***Emily Cross**[1], Oliver Podmanicky, Benjamin Munro, Rita Horvath*
[1]University Of Cambridge, Cambridge, United Kingdom

3.105P Late-onset thymidine kinase 2 deficiency: clinical features and long-term response in adult patients receiving deoxynucleoside therapy
***Hacer Durmus**[1], Asuman Gedikbasi, Arman Cakar, Esen Kıyan, Yesim Parman*
[1]Istanbul Faculty of Medicine, Istanbul University y, Istanbul, Turkey

3.106P Intrafamilial phenotypic variability and variable response to riboflavin therapy in SLC52A2-related riboflavin transporter deficiency: a Turkish family case series
***Ali Zeki Bedir**[1], Haluk Topaloğlu*
[1]Istanbul Medeniyet University / Goztepe Research and Training Hospital, Istanbul, Turkey

Poster Session 3: 3.076eP-3.078eP, 3.107P-3.116P: Vacuolar myopathies

3.076eP Clinicopathological, imaging, and genetic spectrum of inherited rimmed vacuolar myopathies in a Chinese cohort
*Feng Lin, Yu-hua Lin, Ze-ling zheng, Qi-Fang He, Xin Lin, **Xinrong Xu**[1], Kang yang, Ming-hui Zeng, Xin-yue Chen, Min-ting Lin, Ning Wang, Zhi-qiang Wang*
[1]Department Of Neurology Of First Affiliated Hospital, Fujian Medical University, Fuzhou, China

3.077eP Eosinophilia presenting with rhabdomyolysis: case report and literature review

Meirong Liu[1], *Yang Wang, Xiaoke Wu, Na Ji, Qi Fang*
[1]*The First Affiliated Hospital Of Soochow University, Suzhou, China*

3.078eP Bilateral upper-extremity hybrid assistive limb treatment and aceneuramic acid for GNE myopathy

Naoki Suzuki[1], *Hiromitsu Oya, Shun Takanaka, Shunta Yokouchi, Kota Ataka, Shota Soma, Yusuke Sekiguchi, Toshiaki Takahashi, Kensuke Ikeda, Rumiko Izumi, Naoko Nakamura, Midori Miyagi, Takahiro Miura, Tamao Takahashi, Chihiro Nakazawa, Keisuke Obata, Tatsuma Okazaki, Masashi Aoki, Satoru Ebihara*
[1]*Tohoku University, Sendai, Japan*

3.107P X-linked recessive vacuolar myopathy with diabetes and intellectual disability linked to a STK26 variant

Maria Francisca Fajre[1,2], *Sarah Souvannanorath, Gianmarco Severa, Juliette Nectoux, Corinne Metay, Alix De Bec-de-Lievre, Damien Sternberg, Marina Konyukh, Ariane Lunati-Rozie, Benoit Funalot, Edoardo Malfatti*
[1]*University Paris Est Créteil, Inserm, MUSKETHER LAB U955, IMRB, F-94010, Créteil, France,* [2]*Department of Neurology, Hospital San Juan De Dios, Santiago, Chile*

3.108P Natural history and clinical variability in RBCK1-related disease

Ayşe Şenol Ersak, Nagihan Şener, Büşra Hotaman, Damla Yıldız, İlker Ertuğrul, Diclehan Orhan, Gizem Ürel Demir, Kısmet Çıkı, Yılmaz Yıldız, Selman Kesici, Murat Güvener, Deniz Çağdaş Ayvaz, Pelin Özlem Şimşek Kiper, Ali Dursun, ***Göknur Haliloğlu***[1]
[1]*Hacettepe University Faculty of Medicine, Ankara, Turkey*

3.109P Exploratory cognitive and behavioral assessment in ANXA-11 related myopathy

Aldrin Martins[1], *Alberto Rolim Muro Martinez, Anamarli Nucci, Felipe Franco da Graça, Edmar Zanoteli, André Macedo Serafim da Silva, Marcondes Cavalcante França Junior*
[1]*Department of Neurology from University of Campinas, Campinas, Brazil*

3.110P Genetic profile of vacuolar myopathy: retrospective analyses in a single center cohort

Aldrin Martins[1], *Felipe Franco, Alberto Rolim Muro Martinez, Anamarli Nucci, Marcondes Cavalcante França J*
[1]*Neurology Department from University of Campinas, Campinas, Brazil*

3.111P Impact of LAMP2 deficiency on endolysosomal system in Danon disease

Tomo Shiota[1], *Kiichi Nakahira, Minako Yamaoka, Takayuki Shima, Mai Makino, Shuhei Nakamura, Kazuma Sugie*
[1]*Department of Neurology, Nara Medical University, Nara, Japan*

3.112P Characterization of PLIN4 structure and repeat variability in healthy individuals

Sara Testa[1], *Erika Salvi, Cyril Moulin, Sara Gibertini, Lorenzo Maggi, Luc Bousset, Andrey Kajava, Alenka Čopič, Alessandra Ruggieri*
[1]*Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy*

3.113P Decoding the miRNA landscape in PLIN4-related myopathy: from cellular signatures to tissue-level investigation

Eliana Iannibelli[1], *Alessandra Carnazzi, Irene Sambruni, Erika Salvi, Lucia Nicolini De Gaetano, Franco Salerno, Isabella Moroni, Lorenzo Maggi, Alessandra Ruggieri, Sara Gibertini*
[1]*Neurology 4 - Neuroimmunology and Neuromuscular Diseases Unit – Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano, Italy*

3.114P Establishment of a tri-dimensional model for PLIN4-related myopathy using C2C12 stable cell lines

Alessandra Carnazzi[1], *Eliana Iannibelli, Veronica Sian, Jaakko Sarparanta, Per-Harald Jonson, Eleonora Giagnorio, Alenka Čopič, Stefania Marcuzzo, Isabella Moroni, Marco Savarese, Lorenzo Maggi, Sara Gibertini, Alessandra Ruggieri*
[1]*Neuroimmunology and Neuromuscular Diseases Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy*

3.115P Autophagic vacuolar myopathy with AVSF in Japan: an expanded nationwide analysis of Danon disease and XMEA

Kazuma Sugie[1,2,3], *Tomo Shiota, Tomohito Ohashi, Minako Yamaoka, Ai Yamanaka, Rui Shimazaki, Yukako Nishimori, Hitoki Nanaura, Nobuyuki Eura, Takao Kiriyama, Eiichiro Mori, Shuhei Nakamura, Ichizo Nishino*
[1]*Department of Neurology, Nara Medical University, Kashihara, Japan,* [2]*Center for Autophagy and Anti-Aging Research, Nara Medical University, Kashihara, Japan,* [3]*Department of Neuromuscular Research, National Institute of Neurology, National Center of Neurology and Psychiatry, Kodaira, Japan*

3.116P Biomarkers changes in a mouse model of GNE myopathy with 6'-sialyllactose supplementation

Jinhong Shin[1], *Jaeil Choi, Lila Kim, Sumin Kim, Joon Ki Jung, Jaeyoun Bae, Hyeyoung Kim, Sjanie Huang, Dirk Lefeber, Satoru Noguchi*
[1]*Pusan National University Yangsan Hospital, Yangsan, South Korea*

Short Oral Presentations 7: SP28-SP31, Congenital muscle diseases

SP28 A multicentre natural history study in nemaline myopathy in the UK

Gemma Fisher[1], Anna Sarkozy, Giovanni Baranello, Heinz Jungbluth, Maria Vanegas, Meredith James, Andy Yao, Naman Goel, Rui Zhao, Serena Hill, Matthew Woods, Georgia Stimpson, Tina Duong, Sarah Ismail, Carolina Tesi Rocha, Charlotte Lilien, Laurent Servais, Site Study Teams UK, Nemaline Myopathy Data Federation Steering Committee
[1]University Of Oxford UK Neuromuscular Centre, Department of Paediatrics, University of Oxford, Oxford, United Kingdom

SP29 Bulbar dysfunction in CMD-LAMA2: clinical and ultrasonographic assessment

Erick Dupont[1,2], Clara Camelo, Ana Moreira, Mariana Artilheiro, Tatiana Fernandes, Joemir Brito, Pedro Sampaio, Cristiane Moreno, Fernando Kok, Edmar Zanoteli
[1]Universidade De São Paulo - USP, São Paulo, Brazil, [2]Hospital das Clínicas da Faculdade de Medicina de Universidade de São Paulo - HCFMUSP, São Paulo, Brazil

SP30 Defective DNA double-strand break repair through the non-homologous end joining in LMNA-related congenital muscular dystrophy primary myotubes

Marine Leconte, Lucas Ratelet, Zoheir Guesmia, Gisèle Bonne, Anne Bertrand[1]
[1]Myology Research Center, Paris, France

SP31 Phase 1b study of the safety, tolerability, pharmacokinetics, immunogenicity, and efficacy of adimanebart in participants with DOK7-CMS

Hanns Lochmüller, Lorenzo Maggi, Nancy L. Kuntz, Steven J. Burden, Tonke van Bragt, Mitsunori Watanabe[1], Stéphanie Dincq, Willem Talloen, Jamie Lim, Hege Michiels, Sonya Patel, Syed Raza, Kate Lyden, Ieuan Clay, Yaya Zhai, Robert T. Marcotte, Roeland Vanhauwaert, Rebecca Shilling, Jacqueline Palace
[1]argenx, Ghent, Belgium

Short Oral Presentations 8: SP32-SP35, Myotonic dystrophy therapies

SP32 Tricyclo-DNA antisense oligonucleotide compounds to tackle toxic CUGexp-RNA in a mouse model of myotonic dystrophy type 1

Julie Fagioli[1], Cyriaque Beley, Marine Goeffroy, Geneviève Gourdon, Luis Garcia, Denis Furling, Arnaud F. Klein
[1]Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris, France,

SP33 Scaling therapeutic discovery in myotonic dystrophy type 1 through a validated high-throughput screening and machine learning pipeline

Andrea Lopez Martinez[1], Abdullah Al-Ani, Nadia Osegui-Barcenilla, Humberto González-Díaz, Sonia Arrasate, Solange Kapetanovic, Virginia Arechavala-Gomeza
[1]Nucleic Acid Therapeutics for Rare Disorders (NAT-RD), Biobizkaia Health Research Institute, Barakaldo, Spain

SP34 AAV-based gene therapy for myotonic dystrophy

Ludovic Arandel, Aurelien Cordier, Krizia Ronquillo, Samia Martine, Gilles Moulay, Alain Sureau, Antoine Muchir, Michael Blaser, Nathalie Daniele, Isabelle Richard, Arnaud Klein, Denis Furling[1]
[1]Myology Research Center / Institute of Myology, Paris, France

SP35 Preclinical viral and non-viral approaches for myotonic dystrophy gene therapy

Joel Chamberlain[1], Darren Bisset, Stephanie Lee-Diaz, Jinhee Park, Martin Childers, Seung Wook Oh
[1]University of Washington, Seattle, United States

Short Oral Presentations 9: SP36-SP39, 9 Clinical care

SP36 Spectrum of repeat expansion disorders in a neuromuscular cohort in northern India

Farsana Mustafa[1], Ashwin Dalal, Vasantha Sarvade, William L Macken, Lindsay A Wilson, Ayush Agarwal, Robert D S Pitceathly, Rohit Bhatia, ICGNMD Consortium, Michael G Hanna, Kumarasamy Thangaraj K, Venugopalan Y Vishnu
[1]Department of Neurology, All India Institute of medical sciences, New Delhi, New Delhi, India

SP37 Diagnostic journey in patients with Duchenne muscular dystrophy: barriers to early diagnosis in Argentina

Francisco Agustin Cerra[1], Carla Bolaño Díaz, Agustin Jáuregui, Mariel Morosini, Fernando Chloca, José Corderi, Laura Pirra, Gabriel Vázquez, Lilia Mesa, Alberto Dubrovsky
[1]Instituto de Neurociencias del Hospital Universitario Fundación Favaloro, Ciudad Autónoma De Buenos Aires, Argentina

SP38 Facilitating conversations around hope and resilience in progressive neuromuscular disorders

Rebecca Scharf[1], *Dahlia Earleywine, Emily Roth, Anna Jesus, Sarah Jones, Renee Briker, Chelsea Masterson, Leila Orszag, Margaret Plews-Ogan, Erin Plews-Ogan*

[1]*University Of Virginia, Charlottesville, United States*

SP39 The multisystem proteinopathy international multicentre study: preliminary results of an ongoing retrospective data collection

Marianela Schiava[1], *Per Harald Jonson, Ulrika Lindgren, Paloma Martin Jiménez, Lea Leonardis, Andoni Echaniz-Laguna, Beatriz Vélez-Gómez, Daniel Natera-de Benito, Jean-Baptiste Noury, Mário Emílio Dourado, Alvaro Carbayo, Florencia Giliberto, Rosa Elena Escobar Cedillo, Chiara Marini Bettolo, Stephen Wandera, Sonia Segovia, Jordi Diaz Manera - on behalf of the MSP International Study Group*

[1]*John Walton Muscular Dystrophy Research Centre, Newcastle NHS Foundation Trust, Newcastle Upon Tyne, Uk*

Poster Session 4: 4.001eP-4.010eP, 4.001P-4.007P: CMyo - Clinical care and case reports

4.001eP Molecular spectrum and exploratory genotype-phenotype correlations in a prospective natural history cohort of nemaline myopathy

David Zocche[1], *Gemma Fisher, Deirdre Potter, Anna Sarkozy, Giovanni Baranello, Evelin Milev, Meredith James, Emma Robinson, Heinz Jungbluth, Maria Vanegas, Jennie Sheehan, Matthew Woods, Charlotte Lilien, Laurent Servais*
[1]*MDUK Neuromuscular Centre, Department of Paediatrics, University of Oxford, Oxford, United Kingdom*

4.002eP Investigating genetically unresolved cases of congenital fibre-type disproportion by a combination of nanopore sequencing and fibre-type analysis

Ella Nippala[1,2], *Fanny Rostedt, Roosa Harmo, Milla Laarne, Teemu Turku, Harri Kangas, Julien Ochala, Thomas Krag, John Vissing, Manu Jokela, Robert Seaborne, Katarina Pelin, Jenni Laitila*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*Department of Medical Genetics, Medicum, University of Helsinki, Helsinki, Finland*

4.003eP Two boys with X-linked myotubular myopathy presenting with peliosis hepatis after infancy

Minobu Shichiji[1], *Yuki Kihara, Kumiko Ishiguro, Takatoshi Sato, Seisuke Sakamoto, Mureo Kasahara, Keiko Ishigaki*
[1]*Department Of Pediatrics, Tokyo Women's Medical University, School of Medicine, Tokyo, Japan,*

4.004eP Marinesco-Sjögren syndrome: a case report of three siblings due to a novel mutation in the SIL1 gene

Unsal Yilmaz, Selvinaz Edizer, ***Merve Yavuz***[1]
[1]*Gaziantep City Hospital, Turkey*

4.005eP Novel biallelic CRELD1 variants associated with early-onset myopathy and epilepsy

Jorge Alonso-Pérez[1], *Juan Carlos León-Hernández, Marianela Schiava, Leonela Luce, Alejandro González-Chamorro, Abraham Acevedo-Arozena, Olga Tapia, Pablo Rodríguez-García, Nuria Ruiz-Lavilla, Jordi Díaz-Manera, Ana Töpf*
[1]*Neuromuscular Disease Unit. Neurology Department. Hospital Universitario Nuestra Señora de Candelaria. Instituto de Investigación Sanitaria de Canarias (IISC)., Tenerife, Spain*

4.006eP Phenotypic variability in COL12A1-related myopathy: a case series highlighting genotype-phenotype correlations

Sibel Öz Yıldız[1], *Mustafa Gökoğlu, Cengiz Dilber, Pelin Özlem Şimşek Kiper, Gülen Eda Utine, Göknur Haliloğlu*
[1]*Ministry Of Health, Pediatric Neurology Clinic, Necip Fazıl City Hospital, Kahramanmaraş, Turkey,*

4.007eP Risk of acute liver failure in myopathic patients following therapeutic acetaminophen doses: a review of underrecognized risks

Hanneke Dijkhuis, Anne Dittrich, Stephanie Natsch, Saskia de Wildt, ***Hilde Braakman***[1]
[1]*Radboudumc, Nijmegen, Netherlands*

4.008eP Dusty core myopathy related to heterozygous RYR1 gene mutation: description of a first north African family

Emna Farhat[1,2], *Myriam Chaabouni, Najoua Miladi*
[1]*Faculty of Medicine of Tunis, University of Tunis El Manar, 2092., Tunis, Tunisia,* [2]*The Muscle lab, Iris Medical Centre, Hedi Nouira Avenue, Ennasr2, 2037., Tunis, Tunisia*

4.009eP A Korean case of childhood-onset MEGF10-related myopathy with novel compound heterozygous variants

Byeong Joo Choi[1], *Soohyun Kim, Hyung Jun Park, Woo-Kyung Kim*
[1]*Department of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine, Seoul, South Korea*

4.010eP Nemaline cardiomyopathy caused by a missense variant in ACTA1 with a RYR2 variant as a possible genetic modifier

Teemu Turku, Manu Jokela, Milla Laarne, Benedikt Schoser, Hanns Lochmüller, Carina Wallgren-Pettersson, Katarina Pelin, ***Vilma-Lotta Lehtokari***[1,2]
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Faculty of Medicine, Helsinki, Finland*

4.001P Functional trajectories of paediatric patients with recessive TTN-related congenital myopathy – 18 months of prospective natural history data

Charlotte Williams[1,2], *Amy Wolfe, Jennie Sheehan, Alexander Schofield, Maria Vanegas, Gianpaolo Cicala, Jessica Bilby, Teishal Joefield, Francesco Muntoni, Heinz Jungbluth, Giovanni Baranello, Anna Sarkozy*
[1]*Great Ormond Street Hospital for Children, London, United Kingdom,* [2]*Dubowitz Neuromuscular Centre, University College London, London, United Kingdom*

4.002P Novel OBSCN variants linked to recurrent myalgia and severe early onset rhabdomyolysis
Federica Trentin[1,2], *Elena Faedo, Serafina Pacilio, Denise Cassandrini, Penelope Jordan, John Rendu, Alix de Becdelièvre, Marco Savarese, Juliette Nectoux, Edoardo Malfatti, OBSCN study group*
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4.003P Utility of electron microscopy for congenital myopathies: a retrospective review of 425 muscle biopsies
Melissa Mejia-Bautista, Erica Fossee, Steven Moore, ***Osorio Lopes Abath Neto***[1]
[1]*University of Iowa, Iowa City, United States*

4.004P Spatiotemporal patterns of fat replacement in SELENON-related myopathy: a whole-body imaging study
Rui Shimazaki[1,2,3], *Satoru Noguchi, Wakako Yoshioka, Shinichiro Hayashi, Ichizo Nishino*
[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Kodaira, Japan,* [2]*Department of Genome Medicine Development, Medical Genome Center, National Center of Neurology and Psychiatry, Kodaira, Japan,* [3]*Department of Neurology, Nara Medical University School of Medicine, Kashihara, Japan*

4.005P Hidden burden of nemaline myopathy in India: a clinicopathological and genetic exploration
Aishwarya Dhall[1,2], *Bandana Jassal, Venugopalan Y Vishnu, Vaishali Suri, Mehar Chand Sharma*
[1]*Department of Pathology and Lab Medicine, All India Institute of Medical Sciences, Bhubaneshwar, India,* [2]*Neuropathology Laboratory, Neurosciences Centre, All India Institute of Medical Sciences, New Delhi, India*

4.006P The profound impact of X-linked myotubular myopathy across three generations
Lizan Stinissen, Mahalekshmi Desikan, Ros Quinlivan, Sietse Bouma, Baziel G.M. van Engelen, Wija Oortwijn, Kirsten E. Bevelander, ***Nicol Voermans***[1]
[1]*Radboud University Medical Centre, Nijmegen, Netherlands*

4.007P TPM2-related myopathies: enlarging the clinical, genetic and myopathologic spectrum
Denise Cassandrini, Francisca Fajre, Claudio Bruno, Chiara Fiorillo, Jorge Alfredo Bevilacqua, Bornale Das, Marion Onnée, Chiara Panicucci, John Rendu, Giorgio Tasca, Marco Savarese, Paola Tonin, Gaetano Vattemi, Homa Tajshargi, ***Edoardo Malfatti***[1,2]
[1]*University Paris Est Créteil, Inserm, MUSKETHER LAB U955, IMRB, F-94010 , Crétéil, France,* [2]*Reference Center for Neuromuscular Disorders, APHP Henri Mondor University Hospital, Créteil, France*

Poster Session 4: 4.011eP-4.014eP, 4.008P-4.020P: CMyo - Clinical physiopathology

4.011eP In vitro characterisation of mesoporous silica nanoparticles (MSNs) as drug delivery agents in patient-derived models of RYR1-related myopathy
Kara Farstad-O'Halloran[1,2], *Joshua Clayton, Juliana Hamzah, Gianina Ravenscroft, Haritha Kirla, Rhonda Taylor*
[1]*Harry Perkins Institute of Medical Research, Perth, Australia,* [2]*Centre for Medical Research, University of Western Australia, Perth, Australia*

4.012eP Gene replacement therapy for ADSS1 myopathy: preclinical investigation in mice
Kathryn Yammine[1], *Behzad Moghadaszadeh, Emily Hickey, Caroline Lee, Morgan Kim, Jeffrey Brault, Jeffrey Widrick, Alan Beggs*
[1]*Boston Children's Hospital, Boston, United States*

4.013eP AI-driven drug repurposing for ADSS1 myopathy using cross-cohort skeletal muscle transcriptomic signatures
Cara Timpani[1,2], *Priyanka Kakkar, Wenhui Fan, Jiao Kexin, Hyung Jun Park, Emma Rybalka*
[1]*Victoria University, Institute for Health and Sport, St Albans, Australia,* [2]*The University of Melbourne, Department of Medicine – Western Health, Melbourne Medical School, St Albans, Australia*

4.014eP Characterization of adenylosuccinate synthase 1 deficiency in mouse models of inherited myopathy
Dean Campelj[1], *Cara Timpani, Dovile Anderson, Stephanie Kourakis, Ankita Tulangekar, Kathryn Yammine, Behzad Moghadasadeh, Darren Creek, Alan Beggs, Emma Rybalka*
[1]*Institute for Health and Sport, Inherited and Acquired Muscle Diseases Program, Victoria University, Melbourne, Australia*

4.008P Mutant transcript knockdown in RYR1-related myopathy using 2'-O-methyl-modified phosphorothioate antisense oligonucleotides
Anna Johnstone[1,2], *Jordan Crane, Joshua Clayton, Gianina Ravenscroft, Rhonda Taylor*
[1]*Harry Perkins Institute of Medical Research, QEII Medical Centre, Nedlands, WA, Australia, Perth, Australia,* [2]*Centre for Medical Research, University of Western Australia, QEII Medical Centre, Nedlands, WA, Australia, Perth, Australia*

4.009P KBTBD13G67R causes sarcomere dysfunction and Nemaline Myopathy type 6 in patient fibers, but not in mice
Leander Vonk[1], *Rianne Baelde, Alexandra Dorn, Matteo Garibaldi, Josine de Winter, Coen Ottenheijm*
[1]*Amsterdam UMC, Amsterdam, Netherlands*

4.010P Assessing two novel nemaline myopathy mouse models for future therapeutic studies targeting myosin dysfunction

Fanny Rostedt[1,2], *Sonja Heininen, Justin Kolb, Henk Granzier, Jenni Laitila*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*Department of Medical Genetics, University of Helsinki, Helsinki, Finland*

4.011P Ultrastructural correlates of atrophic myofibers in the DNM2 knock-in mouse model of centronuclear myopathy

Genevieve Gucci Grace Uy[1], *Yoshihiko Saito, Tetsuya Takeda, Megumu Ogawa, Hisayoshi Nakamura, Yukiko U. Inoue, Takayoshi Inoue, Ichizo Nishino, Shinichiro Hayashi, Satoru Noguchi*
[1]*Department of Neuromuscular Research, National Center of Neurology and Psychiatry, Kodaira City, Japan*

4.012P Functional validation of aberrant pre-mRNA splicing due to a nonsense variant and exon skipping therapy in MYPN-related congenital myopathy

Guangyu Wang[1], *Chuanzhu Yan, Pengfei Lin*
[1]*Qilu Hospital of Shandong University, Jinan, China*

4.013P Towards improved diagnoses of congenital muscle diseases: an interactive map of non-coding regulatory elements in primary skeletal muscle tissue

Connor McPhan[1,2], *Rhonda Taylor, James Taylor, Joshua Clayton, Matthew Jones, Macsue Jacques, Nir Eynon, Hamid Alinejad-Rokny, Alistair Forrest, Gianina Ravenscroft*
[1]*Harry Perkins Institute of Medical Research, QEII Medical Centre, Perth, Australia,* [2]*Centre for Medical Research, University of Western Australia, Perth, Australia*

4.014P Differential impact of TUBA4A variants on cellular and microtubule morphology in a COS-1 overexpression model

Mridul Johari[1], *Lein Dofash, Jeremy Garcia, Machteld Oud, Ronald van Beek, Gianina Ravenscroft*
[1]*Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia, Nedlands, Perth (Western Australia), Australia*

4.015P Identification of regulatory genomic regions relevant for muscle development as targets for mutation screening in congenital myopathies

Birthe Katrin Alexandra Lange[1,2], *Viktoriia Huryn, Pia Rautenstrauch, Uwe Ohler, Markus Schuelke*
[1]*Charité - Universitätsmedizin Berlin, Berlin, Germany,* [2]*Humboldt-Universität zu Berlin, Berlin, Germany*

4.016P Assessing the potential of iPSC-derived cells to generate 3D skeletal muscle models of ACTA1 disease

Joshua Clayton[1,2], *Vanessa Crossman, Divyashree Avnoor, Sohail Kharrazi, Subash Adhikari, Jeremy Garcia, Christina Vo, Chrystal Tiong, Changho Chun, Darcy Pirotta, Edoardo Malfatti, Norma Romero, David Stroud, David Mack, Nigel Laing, Gianina Ravenscroft, Peter Houweling, Rhonda Taylor*
[1]*Harry Perkins Institute of Medical Research, Perth, Australia,* [2]*University of Western Australia, Perth, Australia*

4.017P Pathogenic mechanisms in Ryr1IT mice, a model of central core disease, and mechanistic insights into miR-486–mediated protective effects

Federica Fiore, Egidio Rubino, Matteo Serano, Annunziatina Laurino, Chiara Ricciardi, Stefano Perni, Enrico Pierantozzi, Daniela Rossi, Martina Lombardi, Stefano Cagnin, Gabriele Sales, Leonardo Nogara, Bert Blaauw, ***Vincenzo Sorrentino***[1,2]
[1]*Department of Molecular and Developmental Medicine, University of Siena, Siena, Italy,* [2]*Azienda Ospedaliero-Universitaria Senese, Siena, Italy*

4.018P Human 3D engineered muscle models of X linked myotubular myopathy enable spatiotemporal dissection of core pathological processes

Valentina Maria Lionello, Lucia Rossi, Leah Zerlin, Daniel Moore, Mariam Zouhair, Owen Li Cheuk Kwan, Eugenia Carraro, Rocco D'Antuono, Kamel Mamchaoui, Anne Bigot, Francesco Muntoni, Andrea Serio, ***Francesco Saverio Tedesco***[1,2,3]
[1]*Department of Cell and Developmental Biology, University College London, London, United Kingdom,* [2]*The Francis Crick Institute, London, United Kingdom,* [3]*Dubowitz Neuromuscular Centre, Great Ormond Street Hospital for Children and UCL GOS Institute of Child Health, London, United Kingdom*

4.019P Building therapeutic understanding in ADSS1 myopathy through multi-omic disease mapping

Emma Rybalka[1,2,3], *Cara Timpani, Khyatiben Pathak, Kamawela Leka, Merve Koç Yekedüz, Raquel van Gool, Jaymin Upadhyay, Wenhui Fan, Jiao Kexin, Hyung Jun Park, Patrick Pirotte*
[1]*Victoria University, Institute for Health and Sport, Melbourne, Australia,* [2]*University of Melbourne, Department of Medicine - Western Health, Melbourne, Australia,* [3]*Cure ADSSL1, Glendale, United States*

4.020P The role of muscle biopsy in the era of genetic testing: essential for diagnosis and clinical decision-making in paediatric myopathies

Aslı Erçili Erdal, Beril Talim, Didem Ardıçlı, Hatice Bektaş Öntaş, Nesibe Gevher Eroğlu Ertuğrul, Ayşegül Neşe Çıtak Kurt, ***Deniz Yüksel***[1]
[1]*University of Health Sciences Turkey, Ankara Etlik City Hospital, Department of Pediatric Neurology, Ankara, Turkey*

Poster Session 4: 4.015eP-4.017eP, 4.021P-4.022P: Congenital myasthenic syndrome

4.015eP The epidemiology of congenital myasthenic syndromes in Japan: an exploration of diagnosed and genetic prevalence
Hirofumi Teranishi[1], *Syed Raza, Xin Zhao, Nicholas Silvestri, Connor Buffel*
[1]*argenx, Ghent, Belgium, 2ISMS, Zoersel, Belgium*

4.016eP Heterozygous DOK7-related congenital myasthenic syndrome responsive to salbutamol: a case series from Iraq
Akram Alnajati[1], *Rasha El Sherif, Ichizo Nishino*
[1]*Al-hadi neuromuscular center, Karbala, Iraq*

4.017eP Epidermolysis bullosa simplex with muscular dystrophy and myasthenia: a case report with plectinopathy
Ercan Demir[1], *Abdullah Sezer, Bülent Cengiz, Gülsüm Kayhan*
[1]*Faculty of Medicine, Department of Pediatric Neurology, Gazi University, Ankara, Turkey*

4.021P CIC-1 chloride channel inhibitor NMD712 improves muscle function in mouse models of congenital myasthenic syndromes
Kelly Ho[1,2], *Ricardo Carmona-Martinez, Martin Skov, Martin Broch-Lips, Thomas Petersen, Sally Spendiff, Hanns Lochmüller*
[1]*CHEO Research Institute, Ottawa, Canada,* [2]*University of Ottawa, Ottawa, Canada*

4.022P TUBA4A pathogenic variant leads to genetic myasthenic syndrome with post-and pre-synaptic defect of neuromuscular transmission
Margherita Milone[1], *Xin-Ming Shen, Zhiyv Niu, Stefan Nicolau, Teerin Liewluck, Eric J Sorenson, C Michael Harper, William J. Litchy*
[1]*Mayo Clinic, Rochester, MN, United States*

Poster Session 4: 4.018eP-4.027eP, 4.023P-4.029P: Muscle imaging

4.018eP Defining MRI-based biomarkers for separating muscle fat neogenesis and atrophy in FSHD
Markus Karlsson, Per Widholm, ***Mary Foltz***[1], *Olof Dahlqvist Leinhard*
[1]*Amra Medical AB, Linköping, Sweden*

4.019eP Muscle imaging features in tubular aggregate myopathies associated with STIM1 and ORAI1 variants
Yukako Yae[1], *Satoru Noguchi, Shinichiro Hayashi, Ichizo Nishino*
[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan*

4.020eP MRI-based assessment of masticatory muscle involvement in hereditary myopathies
Yoshihiko Saito[1], *Wakako Yoshioka, Nobuyuki Eura, Shinichiro Hayashi, Satoru Noguchi, Ichizo Nishino*
[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Kodaira, Japan*

4.021eP High-throughput 3D light-sheet microscopy of intact mouse hindlimbs enables skeletal muscle morphometrics in rodent myopathy models
Max Hahn[1], *Alex Addinsall, Nicolas Eskesen, Anitta Kinga Sárvári, Kasper Andersen, Estrid Pedersen, Marco Tozzi, Henrik H. Hansen, Urmas Roostalu*
[1]*Gubra, Copenhagen, Denmark*

4.022eP Quantitative muscle ultrasound as a new imaging technique: steps toward clinical implementation in Norway
Cecilie Aronsen Haestad[1], *Andreas Rosenberger, Sissel Løseth, Stian Hoven, Kjell Arne Arntzen*
[1]*Norwegian Centre For Rare Diseases, Unit For Neuromuscular Disorders, Tromsoe, Norway,*

4.023eP How useful is muscle MRI in the assessment and differential diagnosis of recessive myotonia congenita (Becker)?
Maren Winkler, ***Jens Reimann***[1]
[1]*Centre for Neurology, University Hospital Bonn, Bonn, Germany*

4.024eP Evaluation of 2-point, 3-point and 6-point Dixon MRI for quantification of muscle fat replacement in patients with muscular dystrophy
Maja Olsen[1], *Mads Godtfeldt Stemmerik, Nanna Scharff Poulsen, Anna Slipsager, Annica Buss Enegaard, Kevin Robert Brow, Adam Espe Hansen, John Vissing*
[1]*Copenhagen Neuromuscular Center (CNMC), Rigshospitalet, Copenhagen, Denmark*

4.025eP Orchidectomy-induced osteosarcopenia in aged rats: a preclinical model for muscle research
Jenni Mäki-Jouppila[1], *Eija Aho, Mari Suominen, Anniina Lehto, ZhiQi Peng, Katja Fagerlund, Jukka Vääräniemi, Jukka Morko, Riikka Oksala, Jukka Rissanen*
[1]*Pharmatest Services, Turku, Finland*

4.026eP Non-invasive imaging techniques for the analysis of muscle, bone, and fat tissues in ovariectomized mice
Jukka Vääräniemi, ***Jenni Mäki-Jouppila***[1], *Yvonne Konkol, Justyna Zdrojewska, Katja Fagerlund, Jukka Rissanen, Jukka Morko*
[1]*Pharmatest Services, Turku, Finland*

4.027eP Atypical skeletal muscle MRI pattern involving the iliopsoas and the short head of the biceps femoris in Stormorken syndrome
Ryoko Inutsuka[1], *Satoshi Morise, Aya Murakami, Wakako Yoshioka, Yasumasa Hashimoto, Ichizo Nishino, Yusuke Yakushiji*
[1]*Department of Neurology, Kansai Medical University, Hirakata city, Osaka, Japan*

4.023P Implementing ultrasound-guided nerve blocks in muscle biopsies in North Norway
Vidar Tor Nyborg Stefansson[1,2], *Tommy Melå Henriksen, Daniel Poppe Skipenes, Christian Enselmann Holgersen, Tine Stokmo, Kjell Arne Arntzen, Linn Hofsøy Steffensen, Andreas Dybesland Rosenberger*
[1]*Norwegian Centre for Rare Diseases - Unit for Neuromuscular Disorders, Tromsø, Norway,* [2]*University Hospital of North Norway, Tromsø, Norway*

4.024P Distinct patterns of muscle involvement among autoantibody subtypes in immune-mediated necrotizing myopathy
Rina Sugie[1], *Nobuyuki Eura, Ai Yamanaka, Rui Shimazaki, Yukako Nishimori, Kazuma Sugie*
[1]*Nare Medical University, Kashihara, Japan*

4.025P What we talk about when we talk about Mercuri: quantitative Dixon fat fraction and Mercuri score concordance in dysferlinopathy muscle MRI
Benjamin Pizarro Galleguillos[1], *Cristian Montalba, José Verdú-Díaz, Felipe Tobar, Jordi Díaz-Manera, Marcelo Andia, Jorge Díaz Jara, Jorge Bevilacqua*
[1]*Imaging Center, University of Chile Clinical Hospital, Santiago, Chile*

4.026P Ranked dissimilarity as a metric for MRI pattern consistency in neuromuscular disorders
Laura Costa Comellas[1,2], *Gabriela Urcuyo, Mireira Álvarez-Molinero, Francina Munell, José Díaz-Verdú, Jordi Díaz-Manera, David Gómez-Andrés, MyoGuide Consortium*
[1]*Hospital Universitari Vall d'Hebrón, Barcelona, Spain,* [2]*Universitat Autonoma Barcelona, Barcelona, Spain*

4.027P Responsiveness to change of quantitative whole-body MRI: results from ReDUX4 OLE
Per Widholm, Markus Karlsson, ***Mary Foltz***[1], *Olof Dahlqvist Leinhard*
[1]*AMRA Medical AB, Linköping, Sweden*

4.028P A clinically feasible relaxation-compensated noninvasive CEST MRI method for reliable detection of skeletal muscle OXPHOS activity
Ritambhar Burman, Lauren Wooten, Samuel Hughes, Asim Bag, Richard Finkel, ***Puneet Bagga***[1]
[1]*Department of Radiology, St. Jude Children's Research Hospital, Memphis, United States*

4.029P A machine learning–based approach to screening neuromuscular disorders using multifrequency electrical impedance myography
Seward Rutkove[1], *Buket Sonbas-Cobb, W. David Arnold, Kaneshia Hives, Amy Bartlett, Peter Riley, Aleah Pagan, Stephen Chrzanowski, Basil Darras, Stephen Kolb*
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Poster Session 4: 4.028eP-4.037eP, 4.030P-4.033P: Myasthenia gravis

4.028eP Statin use preceding myasthenia gravis diagnosis: a nationwide case-control study in Denmark
Josefine Jul Jarbæk Nielsen[1], *Lotte Sahin Levison, Timothy Lee Lash, Henning Andersen*
[1]*Neurology, Aarhus University Hospital, Aarhus, Denmark*

4.029eP Live cell-based diagnostic assay for the detection of low-affinity anti-AChR antibodies in autoimmune myasthenia gravis in France
Arnaud Jacquier[1,2], *julien Carras, Julian Theuriet, Antoine Pégat, Carole Vuillerot, Laurent Schaeffer*
[1]*CNRS UMR5261 INSERM U1315 PGNM, Lyon, France,* [2]*Centre de Biotechnologie Cellulaire, Hospices Civils de Lyon , Bron, France*

4.030eP Clinical and demographic characteristics of patients with very late onset myasthenia gravis
Hamdi Necdet Karli[1], *Emel Oguz Akarsu, Sarra Elhamida Lazrak*
[1]*Bursa Uludag University School of Medicine Dept of Neurology, Bursa, Turkey*

4.031eP A simplified murine model of experimental autoimmune myasthenia gravis (EAMG)
Lukas Theissen[1,2], *Christopher Nelke, Christina Schroeter, Niklas Huntemann, Saskia Räuber, Derya Bachir Bachir, Kathrin Koch-Hölsken, Armin Polzin, Norbert Gerdes, Sven G. Meuth, Tobias Ruck*
[1]*Department of Neurology, BG University Hospital Bergmannsheil, Bochum, Germany,* [2]*BG University Hospital Bergmannsheil, Heimer Institute for Muscle Research, Bochum, Germany*

4.032eP A national study on juvenile myasthenia gravis in Denmark: incidence rate, onset and treatment
Madeleine Rzepa Hopkirk[1], *Josefine Jul Jarbæk Nielsen, Henning Andersen*
[1]*Department Of Neurology, Aarhus University Hospital, Aarhus, Denmark*

4.033eP Clinical spectrum, management and outcomes of myasthenia gravis in Oman: a retrospective observational cohort study in a single tertiary center
Ahmad Elmesallami[1], *Ahmed Nadeem, Ahmed Mohamed, Kawthar Al Salmi, Basma Al Muqaimi, Abdullah Al Salti*
[1]*Khoula Hospital/ Ministry of Health, Muscat, Oman*

4.034eP Very late-onset myasthenia gravis: a single-center experience
Patimat Isabekova[1], *Tatiana Alekseeva*
[1]*Almazov National Medical Research Centre, Saint-petersburg, Russia*

4.035eP Predictors of adverse outcomes in myasthenia gravis: a single-center cohort
Patimat Isabekova[1], *Tatiana Alekseeva*
[1]*Almazov National Medical Research Centre, Saint-Petersburg, Russia*

4.036eP Juvenile myasthenia gravis: an unusual presentation
Patimat Isabekova[1]
[1]*Almazov National Medical Research Centre, Saint-petersburg, Russia*

4.037eP Target antigen switch from CD19 to BCMA in sequential bispecific T-cell engager therapy for refractory myasthenia gravis-Lambert-Eaton overlap syndrome
Niklas Huntemann[1], *Menekse Öztürk, Stefanie Schreiber, Stefanie Lichtenberg, Lars Masanneck, Christopher Nelke, Thomas Ulrych, Dimitrios Mougiakakos, Sascha Dietrich, Sven G. Meuth, Tobias Ruck*
[1]*Department Of Neurology, University Hospital Münster, University Münster, Münster, Germany*

4.030P A national study of thymectomy in juvenile myasthenia gravis
Madeleine Rzepa Hopkirk[1], *Josefine Jul Jarbæk Nielsen, Henning Andersen*
[1]*Department of Neurology, Aarhus University Hospital, Aarhus, Denmark*

4.031P C-terminal agrin fragment as a biomarker in myasthenia gravis
Mathilde Møller Særkjær[1], *Jesper Helbo Storgaard, Tina Parkner, Henning Andersen*
[1]*Aarhus University Hospital, Aarhus, Denmark*

4.032P The effect of surgery on the course of the disease in patients who underwent video-thoracoscopic thymectomy (VATS thymectomy) due to myasthenia gravis
Efsun Kanmaz, Senay Aydin[1], *Ali Murat Akçil, Aylin Canavar, Levent Cansever*
[1]*University of Health Sciences Yedikule Chest Diseases and Thoracic Surgery Training and Research*

4.033P Improving sensitivity of autoantibody testing for myasthenia gravis using cell-based assays
Adrian Budhram[1,2], *Ola Ismail, Kamala Sangam, Lulu Bursztyn, Ario Mirian, Yiu-Chia Chang, Michael Nicolle*
[1]*Western University, London, Canada,* [2]*London Health Sciences Centre, London, Canada*

Poster Session 4: 4.039eP-4.059eP, 4.034P-4.070P: Myotonic disorders and Periodic Paralysis

4.039eP Magnetoencephalography and cognition in myotonic dystrophy type 1
Nikoletta Nikolenko[1], *Arjun Ramaswamy, Enza Leone, Rofael Jabr, Chris Turner, Umesh Vivekananda*
[1]*National Hospital for Neurology and Neurosurgery, University College London Hospitals NHS Foundation Trust, London, United Kingdom*

4.040eP Comparison between long-read genome sequencing technologies and the gold-standard small pool PCR in the study of myotonic dystrophy type 1

Eva Coll Liesa[1], *Alicia López-Martín, Alejandro Montalvà Giménez, Eric Casellas-Sanfelix, Ivàn Pajuelo-Navarro, Clara Moliner, Anna Revert Barberà, Eduard Juanola Mayos, Míriam Almendrote, Sebastián Figueroa-Bonaparte, Luís Orduña, Marc Corral-Juan, Daniel M Borràs, Gisela Nogales-Gadea*
[1]*Germans Trias I Pujol Research Institute (IGTP), Badalona, Spain*

4.041eP Retrospective evaluation of clinical findings, prognosis, electrophysiological studies, and genetic results in patients with myotonia

Neslihan Gunduz, Ayten Gulec, Mehmet Canpolat, Ayşe Caglar Sarilar, Hakan GUMUS, ***Hüseyin Per***[1]
[1]*Erciyes University Department of Pediatric Neurology, Kayseri, Turkey*

4.042eP HARMONIA study design: a global phase 3 trial assessing the efficacy and safety of z-basivarsen in myotonic dystrophy type 1 (DM1)

Aaron Novack[1], *Doug Kerr, Sowmya Charu, Soma Ray, Shuli Yu, Allison Kreuzer, Monica Fay*
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4.043eP Patient-reported experiences, preferences and challenges within palliative and end-of-life care for myotonic dystrophy: a UK community survey

Nikoletta Nikolenko[1], *Emma-Jayne Ashley, Wendy Greenwood*
[1]*University College London Hospitals NHS Foundation Trust, London, United Kingdom*

4.044eP Data from the first cohort of FREEDOM2-DM1 multiple-ascending dose study of PGN-EDODM1 support continued development

Paul Streck[1], *Johanna Hamel, Hanns Lochmuller, Jean-Dennis Brisson, Thurman Wheeler, Jacinda Sampson, Namita Goyal, Jordi Diaz-Manera, Gerald Pfeffer, James Lilleker, Jeffery Statland, Nicholas Johnson, Alberto Aleman, Christopher Turner, Erin O'Ferrall, Brijesh Garg, Gregory Song, Pallavi Lonkar, Stephen Babcock, Jane Larkindale*
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4.045eP Developing a disease progression model for individuals living with myotonic dystrophy type 1

Guillaume Bassez[1], *Valeria A Sansone, Benedikt Schoser, Nicholas E Johnson*
[1]*Institute Of Myology, France*

4.046eP Longitudinal muscle MRI as an outcome measure in myotonic dystrophy type 1: quantitative fat fraction, STIR biomarkers and radiomics

José Verdú Díaz[1,2,3], *Nikoletta Nikolenko, Kieren Hollingsworth, Hanns Lochmüller, Jaume Bacardit, Jordi Diaz-Manera*
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4.047eP The UK myotonic dystrophy patient registry: empowering clinical research and patient voice with an effective translational research tool

Julie Bohill[1], *Aleks Carver, Jassi Sodhi, Chris Turner, Darren Monckton, Channa Hewamadduma, Mark Hamilton, Emma-Jayne Ashley, Michael Walker, Kate Adcock, Chiara Marini-Bettolo*
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4.048eP Clinical and therapeutic evaluation of patients with myotonic disorders: a single-center retrospective study

Fatma Munevver Gokyigit[1], *Busra Elif Yildirim, Rumeysa Tasci Bulca, Mehmet Yalman, Enise Nur Ozlem Tiryaki, Aygul Tantik Pak*
[1]*University of Health Sciences, Gaziosmanpasa Training and Research Hospital, Dept. of Neurology, Istanbul, Turkey*

4.049eP Apathy in myotonic dystrophy type 1: prevalence, subtype differences and clinical correlates

Wander Jacco Adrian Aarts[1], *Lotte L.J. Put, Karlien Mul, Hilde M.H. Braakman, Dirk J.J. Sweere, Annelien A. Duits, Sylvia Klinkenberg, Catherina G. Faber, Isis B.T. Joosten*
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4.051eP Clinical burden of myotonic dystrophy type 1: targeted review of mortality/morbidity and associated multisystemic impacts

Yoshihiko Inuzuka[1], *Ze Cong, Ryan Thompson, Cecilia R F Silva, Megan Watts*
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4.052eP Mitochondrial dysfunction in myotonic dystrophy type 1
Najoua El Boujnouni[1], *Werner Koopman, Roland Brock, Rick Wansink*
[1]*Department of Medical BioSciences, Radboudumc, Nijmegen, Netherlands*

4.053eP Initial MRI and real-life wearable data from a 2-year natural history study of myotonic dystrophy type 1
Julia Gusatovic[1], *Thomas Gürtler, Camilla Thomsen, Stéphane Motola, John Vissing, Nicolai Preisler*
[1]*Copenhagen Neuromuscular Center, Copenhagen University Hospital, Rigshospitalet, Copenhagen, Denmark*

4.054eP Early severe cardiac involvement in myotonic dystrophy type 1: case series
Dong Won Kwack[1], *Dong Wook Kim, Yang-Ki Min, Jeeyoung Oh*
[1]*Konkuk University Medical Center, Seoul, South Korea*

4.055eP Atrial and ventricular pacing proportions in patients with myotonic dystrophy type 1
David Bovenkerk[1,2], *Wander Aarts, Leandre La Fontaine, Lotte Put, Karlien Mul, Kevin Vernooy, Dominik Linz, Catharina Faber, Isis Joosten*
[1]*Department of Cardiology, Cardiovascular Research Institute Maastricht (CARIM), Maastricht University Medical Centre, Maastricht, Netherlands,* [2]*Department of Neurology, Maastricht University Medical Centre (MUMC+), School for Mental Health and Neuroscience, Maastricht, Netherlands*

4.056eP Liver enzyme profiles and fibrosis assessment in myotonic dystrophy type 1: a multicenter observational study
Basil Van Veen[1,2], *Catharina Faber, Daniel Keszthelyi, Karlien Mul, Isis Joosten*
[1]*Department of Neurology and School for Mental Health and Neuroscience, Maastricht University Medical Centre+, Maastricht, Netherlands,* [2]*Department of Gastroenterology and Hepatology, Maastricht University Medical Centre+, Maastricht, Netherlands*

4.057eP Development of the Rasch-built myotonic dystrophy type 2 activity and participation scale (DM2-Activ)
Leandre La Fontaine[1,2], *Manon Damen, Ingemar Merkies, Catharina Faber, Karlien Mul*
[1]*Maastricht University Medical Center+, Maastricht, Netherlands,* [2]*Mental Health and Neuroscience Research Institute, Maastricht, Netherlands*

4.058eP Hyperkalemic periodic paralysis: cross-sectional quantification of muscle fat replacement using MRI
Rebekka Bernhard[1], *Annica Buss Enegaard, Nanna Scharff Poulsen, Maja Olsen, John Vissing, Sonja Holm-Yildiz*
[1]*Copenhagen Neuromuscular Center (CNMC), Copenhagen, Denmark*

4.059eP The effect of progressive resistance training in persons with hypokalemic periodic paralysis
Annica Enegaard[1], *Jeppe Moesgaard, Rebekka Heintze Bernhard, Nanna Scharff Poulsen, Britt Stævnsbo Pedersen, John Vissing, Sonja Holm-Yildiz*
[1]*Copenhagen Neuromuscular Center (CNMC), Copenhagen, Denmark*

4.034P MFM as a follow-up tool in neuromuscular diseases: patient adherence and cooperation in myotonic disorders
Aurelie Barriere[1], *Shams Ribault, Dominique Vincent-Genod, Pascal Rippert, Shotaro Tachibana, Carole Vuillerot, MFM study Group*
[1]*Hospices Civils De Lyon-l'escale, Lyon, France*

4.035P A loss-of-function deep intronic variant in CLCN1 causes autosomal dominant myotonia congenita
Chunyan Cao[1], *Shang Xu, Wanjin Chen, Jiewen Zhang*
[1]*Department of Neurology, Henan Provincial People's Hospital, People's Hospital Of Zhengzhou University, Zhengzhou, China*

4.036P A possible founder effect underlying a homogeneous genetic profile of non-dystrophic myotonia in Chile
Daniela Avila[1], *Marcela Lagos-Lucero, Carmen Vargas-Leal, Carlos Jaque-Almendras, María de los Angeles Beytía-Reyes, Ivonne Zamorano-Valdebenito*
[1]*Pontificia Universidad Católica De Chile, Santiago, Chile*

4.037P Clinical predictors of myotonia permanens in patients with the SCN4A p.Gly1306Glu variant
Daniela Avila[1], *Daniela Regonesi-Polanco, Carlos Jaque-Almendras, Marcela Lagos-Lucero, Oslando Padilla-Pérez, María de los Angeles Beytía-Reyes, Ivonne Zamorano-Valdebenito, Gabriel Bustamente-Muller, Raúl Escobar-Henríquez, Carmen Vargas-Leal, Marne Salazar-Rivera*
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4.038P Safety and efficacy of mexiletine for treatment of paediatric non-dystrophic myotonias: a single centre experience
Ramya Ramesh Babu[1], *DIvesh Pattni, Michael G Hanna, Dipa Jayaseelan, Pinki Munot*
[1]*Dubowitz Neuromuscular Centre and MRC Centre for Neuromuscular Diseases, UCL Great Ormond Street Institute of Child Health, London, United Kingdom*

4.039P Repeat length as a key determinant for disease severity and antisense oligonucleotide activity in myotonic dystrophy type 1

Najoua El Boujnouni[1], *Lise Ripken, Marieke Willemse, Bart van der Sanden, Kornelia Neveling, Alexander Hoischen, Roland Brock, Rick Wansink*

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4.040P Longitudinal muscle-specific miRNAs as biomarkers of disease progression in myotonic dystrophy type 1

Nikoletta Nikolenko[1], *Andrie Koutsoulidou, Kristia Georgiou, Maria Zanti, Kyriaki Michailidou, Marios Tomazou, Tassos C. Kyriakides, Grainne Gorman, Chris Turner, Hanns Lochmüller, Leonidas A. Phylactou*

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4.041P Study of the CGG interruptions in myotonic dystrophy type 1 with long-read genome sequencing technologies

Eva Coll Liesa[1], *Alicia López-Martín, Alejandro Montalvà Giménez, Eric Casellas-Sanfelix, Ivàn Pajuelo-Navarro, Clara Moliner, Anna Revert Barberà, Eduard Juanola Mayos, Míriam Almendrote, Sebastián Figueroa-Bonaparte, Luis Orduña, Marc Corral-Juan, Daniel M Borràs, Gisela Nogales-Gadea*

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4.042P Defining the natural history of myotonic dystrophy type 1 through a nationwide integrated registry

Alba Herrero Gomez[1], *Alvaro S. Larran, Marc Corral-Juan, Clara Moliner Diaz, Anna Revert Barberà, Usua Laresgoiti-Garay, Alejandro Montalva-Gimenez, Alicia López-Martín, Jorge Alonso-Pérez, Solange Kapetanovic, Gerardo Gutiérrez-Gutiérrez, Ana Lara Pelayo, Macarena Cabrera, Andone Sistiaga, Daniel M. Borràs, Sebastián Figueroa-Bonaparte, Virginia Arechavala-Gomeza, Arturo López-Castel, Gisela Nogales-Gadea, DM1-Hub Consortium*

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4.043P Quantitative muscle MRI as a biomarker of disease activity and progression in myotonic dystrophy type 1

Laura Tufano1, Elisabetta Bucci, Vincenzo Solimene, Marta Zerunian, Matteo Garibaldi

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4.044P Non-dystrophic myotonias in focus: genotype–phenotype correlations and outcomes in myotonia congenita from India

Varun Venkat Raghavan[1], *Vykuntaraju K Gowda, Shivaleela Sajjan*

[1]*Department of Medical Genetics, JSS Medical College and Hospital., Mysuru, India*

4.045P Causes of acute death in myotonic dystrophy type 1: a single-center retrospective study from Japan

Yuhei Hasuike[1], *Tsuyoshi Matsumura, Misa Matsui, Naoko Kawamura, Yuiko Watanabe, Tomoko Saito, Motohiro Sudo, Rika Yamashita, Yuki Yonenobu, Tomohiro Yata, Daisuke Nakatsu, Tomomi Furushima, Takuyuki Endo, Akinori Iyama, Makoto Kinoshita, Misaki Yamadera, Kimiko Inoue, Keiko Toyooka, Toshio Saito, Hideki Mochizuki*

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4.046P Fatigue and its associated factors in patients with myotonic dystrophy type 1

Go Kurauchi[1], *Momoko Goto, Kaori Odaira, Hiromi Sato, Shizuko Taima, Seiko Kon, Haruo Fujino, Makiko Endo, Tsuyoshi Matsumura, Masanori P. Takahashi, Hiroto Takada*

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4.047P A single-center retrospective study of the analysis of the cause of death in myotonic dystrophy

Tsuyoshi Matsumura[1], *Yuhei Hasuike, Misa Matsui, Naoko Kawamura, Yuiko Watanabe, Tomoko Saito, Motohiro Sudo, Rika Yamashita, Yuki Yonenobu, Tomohiro Yata, Daisuke Nakatsu, Tomomi Furushima, Takuyuki Endo, Akinori Iyama, Makoto Kinoshita, Misaki Yamadera, Kimiko Inoue, Keiko Toyooka, Toshio Saito, Hideki Mochizuki*

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4.048P On the move: longitudinal insights advancing the understanding of motor function in paediatric myotonic dystrophy type 1

Lynn Orriëns[1], *Renske van der Heijden, Wietske van Dortmont, Lieze Hoogveld, Maaike Pelsma, Lisa Suppers, Saskia Houwen, Hilde Braakman*

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4.049P Brain imaging and neurocognitive features in myotonic dystrophy: a developmental problem or a progressive leukoencephalopathy?

Corrado Angelini[1], *Gabriele Siciliano*

[1]*Campus Pietro d'Abano, Department Neurosciences, Padova, Italy*

4.050P Automated scoring of video hand opening test for grip myotonia using markerless hand tracking and computer vision

Tina Duong[1], *Melina Rogers, Geeta Rajamani, Audrey Chun, Melissa McIntyre, Constance de Monts, Sarah Ismail, Shelby Vogt-Domke, John Day*

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4.051P Impact of assisted reproductive technologies on reproductive outcomes in women with myotonic dystrophy: a retrospective study

Patricia Garay Albizuri[1,2,3], *Roberto Fernández Torrón, Pablo Iruzubieta, Juan José Poza, Maria José Iñarra Velasco, María Lure Berregui, Loreto Martorell, Adolfo López de Munain*

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4.052P Toward personalized cardiac risk stratification in DM1: the biomarker-driven DM1-heart project

Gisela Nogales-Gadea[1], *Anna Revert-Barberà, Eduard Juanola-Mayos, Ainhoa Sancho-García, Victoria Delgado, Martín Rojas-Goldsack, Bernat Bertran-Recasens, Alba Ramos-Fransi, Laura Llansó, Teresa Casadevall, Jorge Espinosa, Maria Salvadó, Velina Nedkova-Hristova, Nicolau Guanyabens, David Reyes-Leiva, Nuria Cayuela-Caudevilla, Javier Sotoca, Alicia López-Martín, Eva Coll Liesa, Daniel M. Borràs*

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4.053P Study for adipose tissue insulin resistance in myotonic dystrophy type 1

Hiroto Takada[1], *Seiko Kon, Yoshibnobu Oyama, Tamaki Kimura, Yasuhito Wakasaya*

[1]*Aomori National Hospital, Aomori, Japan*

4.054P A wake-up call: respiratory dysfunction in children with myotonic dystrophy type 1

Lynn Orriens, Seline van der Kop, Haddia Ghafuri, Peter Merkus, Sylvia Klinkenberg, ***Hilde Braakman***[1]

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4.055P Changes in physical function outcomes in a Japanese cohort of patients with myotonic dystrophy: results from a 3-year natural history study

Makiko Endo[1], *Harumasa Nakamura, Tsuyoshi Matsumura, Hiroto Takada, Satoshi Kuru, Michio Kobayashi, Akinori Nakamura, Masafumi Matsuo, Kazuma Sugie, Takashi Kimura, Haruo Fujino, Hisateru Tachimori, Masanori P. Takahashi*

[1]*National Center of Neurology And Psychiatry, Tokyo, Japan*

4.056P Study for well-being and psychological distress in caregivers of patients with myotonic dystrophy type 1

Momoko Goto[1], *Kaori Odaira, Go Kurauchi, Shizuko Taima, Asami Akasaka, Seiko Kon, Haruo Fujino, Makiko Endo, Tsuyoshi Matsumura, Masanori P Takahashi, Hiroto Takada*

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4.057P Characterizing brain MRI finding in pediatric myotonic dystrophy: a single institution experience

Meghan Moore Burk[1], *Caroline Meyer, Carolyn Kelley, Paige Naylor, Susan Apkon, Michele Yang, Melissa Wright*

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4.058P Patient priorities for treatment outcomes in myotonic dystrophy type 1: a mixed-methods study

Isis Joosten[1], *Basil van Veen, Leandre la Fontaine, Lotte Put, Maurice Sopacua, Johanna Bruijnes, Jorg van Gent, Bas Haasakker, Karlien Mul, Catharina Faber*

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4.059P Natural history of PR interval and QRS duration in myotonic dystrophy type 1

David Bovenkerk[1,2], *Wander Aarts, Leandre La Fontaine, Sander Van Kuijk, Lotte Put, Karlien Mul, Kevin Vernooy, Dominik Linz, Catharina Faber, Isis Joosten*

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4.060P Comprehensive natural history in children with myotonic dystrophy: a single-institution experience over 20 years

Meghan Moore Burk[1], *Caroline Meyer, Carolyn Kelley, Paige Naylor, Susan Apkon, Michele Yang*

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4.061P Evaluation of ambulatory function with a wearable sensor in myotonic dystrophy type 1

Margaux Poleur, Stéphanie Delstanche, Manon Hustinx, Guillaume Parinello, Noor Benmhammed, Camille Bisson, Charline Dubois, Manon Duclos, Thao Nguyen Le Lam, Laurie Médard, ***Damien Eggenspieler***[1], *Laurent Servais*

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4.062P Integrating clinical, functional, cognitive, and molecular phenotypes to advance precision medicine in myotonic dystrophy type 1

Clara Moliner Diaz[1], *Alba Herrero-Gómez, Alvaro S. Larran, Marc Corral-Juan, Daniel M. Borras, Anna Revert Barberà, Luisa Tenorio, Álvaro Gómez-Ferrer Ardid, Cynthia Almeida, Marina Gonzalez Alvarez, Bibiana Chinea Jimenez, Estefanía Bocanegra, Mireia Lozano Royo, Irati Larrañaga, Patricia Garay, Andone Sistiaga, Jorge Alonso-Pérez, Arturo López-Castel, Gisela Nogales Gadea, DM1-Hub Consortium*

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4.063P Adaptive functioning and caregiver burden in congenital and childhood myotonic dystrophy: a nationwide survey in Japan

Haruo Fujino[1], *Yuko Shimizu-Motohashi, Minobu Shichiji, Rie Honda, Michio Kobayashi, Tsuyoshi Matsumura, Yuzuha Ichimura, Atsuko Ishii, Yasuhiro Takeshima, Harumasa Nakamura, Midori Senoo, Hirofumi Komaki, Keiko Ishigaki, Masanori P. Takahashi*

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4.064P Muscle strength and outcome measures for upper limb function in adults with myotonic dystrophy type 1: a cross-sectional study

Hanne Ludt Fossmo, Hilde Stendal Robinson, Karianne Grüner Bjåstad, Kristin Nielsen Varhaug, Jan Frich, ***Kristin Ørstavik***[1]

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4.065P Preimplantation genetic testing in myotonic dystrophy type 1: clinical outcomes and insights

Johanna Bruijnes[1], *Fauve van Veen, Malou Heijligers, Ronald van Golde, Marianne van Buul-van Zwet, Alwin Derijck, Nicole Corsten-Janssen, Catharina Faber, Els Vanhoutte, Isis Joosten*

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4.066P Gastrointestinal involvement in myotonic dystrophy type 1: an integrated retrospective analysis

Basil Van Veen[1,2], *Isis Joosten, Catharina Faber, Daniel Keszthelyi, Wander Aarts, Karlien Mul, José Conchillo*

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4.067P Functional confirmation of KCNA7 as a new disease gene for hypokalemic periodic paralysis

Gourav Saha, Khurram Liaqat, Kayla Treat, Summan Thahiem, John Kincaid, Fenfen Wu, Francesco Vetrini, Erin Conboy, ***Steve Cannon***[1]

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4.068P Hypokalemic periodic paralysis meets panhypopituitarism: a novel phenotype

Paula Panos-Basterra, Oceana Vieira Pinto-Delaville, Guillaume Taieb, Jean Baptiste Noury, Damien Sternberg, Christel Jublanc, ***Rocio Nur Villar-Quiles***[1,2,3], *Savine Vicart*

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4.069P Identification and targeted sequencing of spliceopathy biomarkers in the vastus lateralis of myotonic dystrophy type 1 patients

Mark Tarnopolsky[1], *Donald Xhuti, Sumon Datta, Greg Miller, Joshua Nederveen, Peter Shepard, Alessandra Chiarot, Darragh Buckley, Doane Chilcoat*

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4.070P Role of cardiac MRI in detecting silent myocardial changes in myotonic dystrophy type 1

Sebastián Ariel Figueroa Bonaparte[1,2], *Eduard Juanola-Mayos, Anna Revert, Ainhoa Sancho, Martín Rojas-Goldsack, Victoria Delgado, Albert Teis, Javier Sotoca, Raúl Juntas-Morales, Maria Salvado-Figueras, Bernat Bertran Recasens, Alba Ramos-Fransi, Laura Llansó, Jorge Espinosa-García, Teresa Casadevall-Codina, Velina Nedkova-Hristova, Antonio Cano, David Reyes-Leiva, Daniel Borràs, Gisela Nogales-Gadea*

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Poster Session 4: 4.060eP-4.088eP-4.071P-4.092P: Registries, networks and care

4.060eP Understanding barriers to participate in specialized neuromuscular rehabilitation experienced by people with neuromuscular diseases

Charlotte Handberg[1,2], *Ulla Werlauff*

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4.061eP Exploring patient and research site perspectives on the implementation of home nursing in Duchenne muscular dystrophy clinical trial protocols

Emma Heslop[1], *Megan McNiff, Michela Guglieri*
[1]*Jwmdrc, Newcastle University, Newcastle, United Kingdom*

4.062eP Diagnostic journeys in genetic muscle disorders: a scoping review of delays and barriers to genetic diagnosis

Lucy Hickson[1], *Nawab Ali, Grace McMacken, Amy Jayne McKnight*
[1]*Centre for Public Health, Queen's University Belfast, Belfast, United Kingdom*

4.063eP Impact of local neuromuscular clinic access on patient travel distance, costs, and work loss: A case for a community clinic's role in multidisciplinary care

Elizabeth Lynne Wood[1], *Susan Apkon*
[1]*Billings Clinic, Billings, MT, United States*

4.064eP The UK facioscapulohumeral muscular dystrophy patient registry: a powerful tool to support clinical research and patient voice in the translational research pathway

Julie Bohill[1], *Aleks Carver, Robert Muni-Lofra, Andrew Graham, Fiona Norwood, Mark Roberts, Tracey Willis, Emma Matthews, Mark McQueen-Mencias, Kate Adcock, Enrico Bugiardini, Giorgio Tasca, Chiara Marini-Bettolo*
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4.065eP Transitioning from pediatric to adult care in neuromuscular diseases: a management model in the Tuscany region, Italy

Sara Loprieno[1], *Francesca Torri, Michele Sacchini, Chiara Ticci, Guya Astrea, Bianca Buchignani, Mariaconcetta Rende, Roberta Battini, Elena Procopio, Gabriele Siciliano, Giulia Ricci*
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4.066eP ERN EURO-NMD AI PathologyHub: Supporting the biological classification of H&E-stained muscle biopsy slides in neuromuscular disease

Ahmed Abdelrahman, E RN EURO-NMD AI Pathology Hub study group, Gianmarco Severa, ***Edoardo Malfatti***[1,2,3]
[1]*Université Paris Est Créteil, Inserm, U955, IMRB, F-94010 Créteil, France, Crétéil, France,* [2]*APHP, Centre de Référence des Maladies Neuromusculaires, Hôpital Universitaire Henri Mondor, Créteil, France, Crétéil, France,* [3]*ERN EURO-NMD, Paris, France*

4.067eP The LGMD2A/calpainopathy registry: a patient-powered natural history study and trial recruitment tool

Jennifer Levy[1], *Annie Poll, Jordan Boslego, Michela Guglieri, Ann Martin, Katherine Mathews, Michele Wrubel*
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4.068eP From fragmentation to collaboration: building a multidisciplinary neuromuscular ecosystem in India

Himshweta Balwani[1,2,3], *Sneha Shyamprasad, Gauri Khandekar, Sharanya Ravindra, Saravanan Palaniappan, Madhuri Maganthi, Ilin Kinimi, Radhika Manohar, Vinitha Varghese, Aneesh Kumar, Mahima Paul, Gauri Krishna, Thabit Ahmed, Suma Uday, Malathi Voora, Ann Mathew*
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4.069eP Evaluation of trunk control in neuromuscular diseases: a systematic review of measurement properties of trunk-specific instruments

Bayram Sırrı[1], *Fatma Uğur, Numan Bulut, Aysu Kahraman, İpek Gürbüz, Öznur Tunca*
[1]*Muş Alparslan University, Muş, Turkey*

4.070eP Relative patient burden in patient-reported outcomes for neuromuscular clinical trials: incorporating relevance and ease of completion beyond time alone

Brett Kinsler[1], *Jennifer Weinstein, Preshetha Kanagaiah, Chad Heatwole*
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4.071eP Educational booklet for health management of caregivers of muscular dystrophy patients in Japan

Masatoshi Ishizaki[1], *Yuishi Aizawa, Michio Kobayashi, Haruo Fujino, Miku Yamaguchi, Akiko Wakisaka, Tsuyoshi Matsumura*
[1]*Department of Neurology, National Hospital Organization Kumamoto Saishun Medical Center, Kumamoto City, Japan*

4.072eP Leveraging partnerships between small and large care centers to provide telehealth consultation improves access and equity

Elizabeth Lynne Wood[1], *Susan Apkon*
[1]*Billings Clinic, Billings, MT, United States*

4.073eP Diagnostic odyssey and its impact on caregivers in a progressive neuromuscular disorder: a qualitative study with quantitative insights

Sneha Banavara Shyamprasad[1], *Himshweta Balwani, Gauri Khandekar, Sharanya P Ravindra, Ann A Mathew*
[1]*Synapse Neuro Center, Bengaluru, India*

4.074eP Lifestyle interventions in muscle disease: a systematic review of exercise and dietary programs in myopathies

Esther Brusse[1], *Gamida Ismailova, Quinten Vervaart, Margreet Wagenmakers, Linda Scheffers, Koen Dekkers, Eric Voorn, Ans Van der Ploeg, Linda Van den Berg*
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4.075eP A multi-disciplinary education programme for healthcare professionals in care and research for Duchenne muscular dystrophy: Duchenne education in care and research initiative (DECRI)

Catherine Turner[1], *Emma Heslop, Adam Kerr, Megan McNiff, Michela Guglieri*
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4.076eP From user centred design to participatory co-design in neuromuscular assistive technologies

Michael Bond[1], *Hayley Philippault, David Buckley, Theofilos Kempapidis, Oisin Minogue, Cara Shaw, Benjamin James, Hamane Sfalsin, Alexandra Johnson, Emily Reuben, Farnaz Nickpour*
[1]*Duchenne UK, London*

4.077eP The promotion of health and employment support for the patients with muscular dystrophies

Takahiro Nakayama[1], *Shizuka Katagiri, Miyae Ogawa, Haruna Sugawara*
[1]*Yokohama Rosai Hospital, Yokohama, Japan*

4.078eP Global FKRP registry: data trends and highlights from over 1,000 participants

Julie Bohill[1], *Jessica Moraes, Lindsay Alfano, Kelly Brazzo, Nicholas Johnson, Jean-Pierre Laurent, Katherine Mathews, Simone Thiele, John Vissing, Maggie Walter, Lacey Woods, Kristin Ørstavik, Volker Straub*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle Upon Tyne, United Kingdom*

4.079eP TREAT-NMD Global registry network: an insight into a large global neuromuscular patient dataset

Miriam Rodrigues[1,2], *Anna Baines, Annie Poll, Neil Bennett, Michela Guglieri, . TREAT-NMD Global Registry Network*
[1]*Auckland City Hospital, Auckland, New Zealand,* [2]*University of Auckland, Auckland, New Zealand,*

4.080eP Myotubular and centronuclear myopathy: analysis of registry data to understand current nutritional practices and explore potential associations between liver health and nutrition

Julie Bohill[1], *Anne Lennox, Erin Ward, Orla Lynch, Alex Roca, Ulrike Schara-Schmidt, Robert Graham, Edmar Zanotelli, Ana Buj-Bello, James Dowling, Heinz Jungbluth, Chiara Marini-Bettolo*
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4.081eP PaLaDIn – improving decision making in neuromuscular diseases (NMDs). What are the benefits of using the interactium data platform?

Rebecca Leary[1], *Emma Heslop, Volker Straub, Alison Bell, Neil Bennett, David Allison, Ilaria Zito, Bouchra Ezzamouri, Marco Roos, Martijn Kersloot, Benedikt Schoser, Amanda Hill, Mireia Claramunt, Luis Querol*
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4.082eP Duchenne UK connect: a digital patient platform and app supporting trial readiness, research participation and care planning in Duchenne muscular dystrophy

Michael Bond[1], *Bouchra Ezzamouri, Emma Heslop, Hayley Philippault, Phillip Cammish, Catherine Turner, Alexandra Johnson, Emily Reuben, Volker Straub, Michela Guglieri*
[1]*Duchenne UK, London, United Kingdom*

4.083eP Webinar on the health of caregivers for patients with muscular dystrophy

Michio Kobayashi[1], *Masatoshi Ishizaki, Tsuyoshi Matsumura*
[1]*NHO Akita National Hospital, Yurihonjo, Japan*

4.084eP HERCULES use of lived experience to inform a disability-based model of the cost of Duchenne muscular dystrophy (DMD)

Emily Reuben OBE[1], *Fleur Chandler, Josie Godfrey, Ros Quinlivan, Jon Hastie*
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4.085eP Long-term survival, quality-adjusted life-years, and economic burden in limb-girdle muscular dystrophy type 2I/R9: a health outcomes model

*Chet Villa, Lucille Sun, Karen Bartley, Dimah Saadé, Michelle Staron-Ehlinger, Anna Talaga, David Veenstra, Lisa Bloudek, **Sajin Mathew**[1], Katherine Mathews*
[1]BridgeBio Pharma, Inc, Palo Alto, United States

4.086eP "Neuromuscular disorders in a nutshell": an innovative way of visualizing clinical summaries for rare neuromuscular diseases

Geir Hevnskjel Ringvold[1], *Andreas D. Rosenberger, Kjell Arne Arntzen, Tine Stokmo*
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4.087eP FSHD Europe: Strengthening the European FSHD ecosystem: bridging clinical care, research, and patient-centered outcomes

*Ria de Haas, Sheila Hawkins, **Nicol Voermans**[1]*
[1]Radboud University Medical Centre, Nijmegen, Netherlands

4.088eP Sexual health in neuromuscular disease: knowledge, attitudes and practices of a multidisciplinary team: the E.M.B.R.A.C.E. study

Jacopo Luca Casiraghi[1], *Maria Beretta, Susanna Pozzi, Bradley Hattingh, Carola Ferrari Aggradi, Alice Zanolini, Agliuzzo Claudia, Irene Malberti, Valeria Ada Sansone*
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4.071P Exclusion, deficit framing, and reduced patient agency in disability-related sexual health research: a triple burden revealed by a computational analysis

Audrey El Kaim[1], *Frederic Fer, Jean-Yves Hogrel, Marcela Gargiulo*
[1]Association Institute of Myology, Paris, France

4.072P Walking on – the ambulate NMD project tracking longitudinal gait function in childhood-onset neuromuscular disease

Kate Carroll[1,2], *Katy de Valle, Justine Adams, Andrew Kornberg, Ian Woodcock, Eppie Yiu, Rachel Kennedy*
[1]The Royal Children's Hospital, Melbourne, Australia, [2]Murdoch Children's Research Institute, Melbourne, Australia

4.073P Concurrent and cross-contextual validity of the functional mobility scale as a patient reported outcome measure in childhood-onset neuromuscular disease

Rachel Kennedy[1,2], *Jeniffer Moon, Adrienne Harvey, Leon Di Stefano, Justine Adams, Katy de Valle, Kate Carroll*
[1]Murdoch Children's Research Institute, Parkville, Afghanistan, [2]The Royal Children's Hospital, Parkville, Australia

4.074P NM-score large-scale validation in 1281 patients with various NMDs: a useful classification system for different clinical presentation

Shotaro Tachibana[1,2], *Shams Ribault, Dominique Vincent-Genod, Pascal Rippert, Aurélie Barrière, Carole Vuillerot, MFM Study Group*
[1]Hospices Civils De Lyon - Escale, Lyon, France, [2]Lyon Neuroscience Research Centre - Trajectoires, Bron, France

4.075P Normative reference data for axial and handgrip muscle strength in healthy adults

Thomas Cortez Gürtler[1], *Karen S Rudolf, Aisha M Sheikh, Julia Gusatovic, Nanna Witting, John Vissing*
[1]Copenhagen Neuromuscular Center, Copenhagen, Denmark

4.076P A population-based study among Swedish adults with non-congenital muscular dystrophies with focus on prevalence and genotypic spectrum

Sara Elisabeth Nordström[1,2], *Carola Hedberg-Oldfors, Lindgren Ulrika, Lisa Wahlgren, Anders Oldfors, Christopher Lindberg, Kalliopi Sofou*
[1]Department of Neurology, Sahlgrenska University Hospital, Gothenburg, Sweden, [2]Department of Paediatrics, Institute of Clinical Sciences, Sahlgrenska Academy, University of Gothenburg, Gothenburg, Sweden

4.077P Tracking disease in motion: wearable-derived insights in neuromuscular disorders

Jean-yves Hogrel[1], *Thomas Marques, Pauline Santmarty, Valérie Decostre, Robert Muni-Lofra, Volker Straub, The DT4RD Consortium*
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4.078P IMU-based workspace metrics for longitudinal assessment of upper limb function in Duchenne muscular dystrophy and spinal muscular atrophy

*Alessandra Favata, Jesica Expósito-Escudero, Roger Gallart-Agut, Julita Medina-Cantillo, Carme Torras, Josep M Font-Llagunes, Rosa Pàmies-Vilà, **Daniel Natera De Benito**[1]*
[1]Hospital Sant Joan De Deu, Barcelona, Spain

4.079P Serum myostatin as a diagnostic biomarker in neuromuscular disorders: evaluation in a large cohort and correlation with clinical surrogates of disease severity

Klara Antonia Trusch, David Strube, Caroline Deborah Stapf, Thomas Kendzierski, Anna Katharina Koelsch, Laura Foerster, Tobias Mayer, Vera-Elena Kendzierski, Caroline Kulitze, Ilka Schneider, Lorenzo Barba, Arthur Guthknecht, Markus Otto, Petra Steinacker, ***Alexander Mensch***[1,2]

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4.080P Toward accessible gait assessment: validity and reliability of a monocular markerless approach in neuromuscular diseases

Pauline Santmarty[1,2]*, Thomas Marques, Frédéric Fer, Guillaume Bassez, Jean-Yves Hogrel, Coralie Bertrand, Hélène Pillet, Laurent Gajny, Romain Feigean*

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4.081P Development and content validity of a trunk-specific assessment scale for children with neuromuscular disorders

Bayram Sırrı[1]*, Numan Bulut, Aysu Kahraman, İpek Gürbüz, Öznur Tunca*

[1]*Muş Alparslan University, Muş, Turkey*

4.082P Caregiver impact in neuromuscular conditions: an Indian perspective

Sneha Banavara Shyamprasad[1]*, Himshweta Balwani, Gauri Khandekar, Sharanya P Ravindra, Ann A Mathew*

[1]*Synapse Neuro Center, Bengaluru, India*

4.083P Global registry for COL6-related dystrophies: improving data collection and harmonisation

Julie Bohill[1]*, Jessica Moraes, Valérie Allamand, Rachel Alvarez, Harry Boddy, John Copier, Nicolas Deconinck, Gustavo Dziewczapolski, Xavi Ferré, Bruce McAlister, Alexandre Mejat, Anna Sarkozy, Volker Straub*

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4.084P Standardized respiratory care for pediatric neuromuscular disorders in the intensive care unit

Wan-Ling Hsiao[1]*, Wen-Chen Liang*

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4.085P Improving healthcare decision making in Duchenne muscular dystrophy through nationwide health technology assessment by using real-world data

Max Duintjer[1]*, Yvonne Meijer-Krom, Rianne Hoek, Dennis Mook-Kanamori, Wilbert van den Hout, Saskia Houwen, Erik Niks, Femke Klouwer*

[1]*Leiden University Medical Center, Leiden, Netherlands*

4.086P Understanding adherence to home-based exercise in youth with neuromuscular disorders: a scoping review

Allison Grangé[1,2]*, Jennifer Boulais, Maryam Oskoui, Danielle Levac*

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4.087P Successfully matching patients with specialized physiotherapists – results from a Norwegian innovation project

Cathrine Ramberg[1]*, Geir Hevnskjel Ringvold, Tollef Ladehaug, Simen Stokke, Kristin Ørstavik, Andreas Dybesland Rosenberger*

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4.088P A multi-stakeholder communication strategy to accelerate recruitment and engagement in the DM1-Hub, the myotonic dystrophy type 1 natural history registry

Usue Laresgoiti Garay, ***Alvaro S. Larran***[1]*, Alba Herrero Gómez, Marc Corral-Juan, Daniel Borras, Clara Moliner, Luis Orduña, Gerardo Gutiérrez-Gutiérrez, Luisa Tenorio, Bibiana Chinea Jimenez, Javier Baigorri, Virginia Arechavala-Gomeza, Arturo López-Castel, Gisela Nogales-Gadea*

[1]*Germans Trias I Pujol Research Institute (IGTP, Spain)*

4.089P Co-designing advance care planning with young adults with NMD: identifying gaps in trust, transparency and therapeutic understanding

Kathryn Irving[1]*, Marnie Graco*

[1]*Royal Children's Hospital, Melbourne, Australia*

4.090P Hereditary neuromuscular disorders in Estonia: a nationwide clinical and genetic overview

Siiri Sarv[1,2], *Tiina Kahre, Eve Oiglane-Shlik, Liis Vali, Katrin Ounap*

[1]*Institute of Clinical Medicine, Faculty of Medicine, University of Tartu, Tartu, Estonia,* [2]*Genetics and Personalized Medicine Clinic, Tartu University Hospital, Tartu, Estonia*

4.091P The CoMPaSS-NMD neuromuscular genome atlas: a new AI-based platform for advanced deep phenotyping and stratification in hereditary neuromuscular disorders

Andi Nuredini[1], *Riccardo Corrias, Daniel Mendelsohn, Sara Pini, Ainara Garzo Manzanares, Michael Obach, Noemi Albano, Marek Socha, Michal Marczyk, Jocelyn Laporte, Ana Topf, Marcello Paolo Scipioni, Jordi Diaz Manera, Volker Straub, Marco Savarese, Joanna Polanska, Filippo Maria Santorelli, Benedikt Schoser, Rossella Tupler, CoMPaSS-NMD Consortium*

[1]*University of Modena and Reggio Emilia, Modena, Italy*

4.092P Perspectives of parents on SMA and DMD diagnosis and early treatment following newborn screening: a phenomenological study

Homira Osman, Lisa Anne Rasmussen, Hugh McMillan, ***Maryam Oskoui***[1,2]

[1]*Departments of Pediatrics and Neurology and Neurosurgery, McGill University, Montreal, Canada,* [2]*Research Institute of the McGill University Health Centre, Montreal, Canada*

Short Oral Presentations 10: SP40-SP43, Neurogenic disorders

SP40 Refining the clinical characterization of PIEZO2-related disorders

Giuliana Capece[1], *Maria Francesca Di Feo, Ali Oghabian, Bjarne Udd, Marco Savarese, PIEZO2 study group*

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SP41 Prenatal intervention in an SMA mouse model rescues neurodevelopmental brain defects associated with spinal muscular atrophy

Charalambos Demetriou[1,2], *Karl Frontzek, Annika Arora, Chaobo Wang, Qiang Zhang, Sachin Deshpande, Mariacristina Scoto, Francesco Muntoni, Haiyan Zhou, Naresh Hanchate, Giovanni Baranello*

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SP42 PACSIN3 mutations impair sensory neurons: insights from patient-derived and CRISPR knock-in models

Shams Ribault[1,2], *Orlane Cardot, Perrine Devic, Valérie Risson, Julien Carras, Laurent Coudert, Julian Theuriet, Philippe Latour, Isabelle Quadrio, Carole Vuillerot, Hélène Puccio, Laurent Schaeffer, Arnaud Jacquier*

[1]*Université Claude Bernard Lyon 1, Lyon, France,* [2]*Hospices Civils de Lyon, Lyon, France*

SP43 Preclinical evaluation of AAV knockdown-and-replace gene therapy in CMT1B mouse models and a humanized peripheral nerve organoid platform

Mary McCulloch[1,2], *Jingting Zhu, Daniella Munezero, Arun Paripati, Noushin Saljoughian, Gongbo Guo, Stefan Nicolau, Nizar Saad, Mark Hester, Afrooz Rashnonejad*

[1]*Jerry R Mendell Center for Gene Therapy, The Abigail Wexner Research Institute at Nationwide Children's Hospital, Columbus, United States,* [2]*Molecular, Cellular, and Developmental Biology Program, The Ohio State University, Columbus, United States*

Short Oral Presentations 11: SP44-SP48, LGMD

SP44 Advancing the understanding of clinical phenotypes using standardized outcome assessments: Insights from the GRASP consortium

Megan Iammarino[1], *Michael Kiefer, Stacy Dixon, Doris Leung, Linda Lowes, Peter Kang, Katherine Mathews, Tahseen Mozaffar, Jeffrey Statland, Conrad Weihl, Matthew Wicklund, Jennifer Raymond, Ruby Langeslay, Lindsay Alfano, Nicholas Johnson*
[1]*University of Texas Health San Antonio, San Antonio, United States*

SP45 SNUPN-related muscular dystrophy (LGMDR29) is a loss-of-function disease with a variable phenotype

Pablo Iruzubieta[1,2], *Alberto Damborenea, Mihaela Ioghen, Nuria Muelas, Katharina Vill, Laura Pérez-Fernández, Ana Töpf, Saioa Moragón, Simon Bajew, Roberto Fernández-Torrón, Gorka Gereñu, Sonia Alonso-Martin, Volker Straub, David de Sancho, Raluca Teleanu, Adolfo López de Munain, Juan J Vílchez, Lorea Blazquez, SNUPN-study group Study group*
[1]*Mcgill University, Montreal, Canada,* [2]*Biogipuzkoa HRI, Donostia - San Sebastian, Spain*

SP46 The extracellular matrix as a driver of muscle degeneration in sarcoglycanopathies: a human proteomic study

James Clark, Esther Fernandez-Simon, Rasya Gokul-Nath, Panos Katsikis, Chiara Panicucci, Serena Baratto, Antonella Pini, Flavia Palombo, Aurelio Hernandez-Lain, Andrew Galloway, Dan Cox, Pawel Palmowski, Andrew Porter, Yolande Parkhurst, Cristina Dominguez-Gonzalez, Maria Lucia Valentino, Claudio Bruno, ***Jordi Diaz-Manera***[1]
[1]*Newcastle University, Newcastle, United Kingdom*

SP47 Engineered extracellular vesicles deliver dysferlin transcripts to dysferlin-deficient myoblast cells to enhance plasma membrane repair

Gianni Giarrano[1], *Mia Kordowski, Juan David Salazar-Gil, Carman Taylor, Vidhi Amin, Caden Islam, Daniel Velásquez-Ramírez, Britani Blackstone, Heather Powell, Christoph Lepper, Noah Weisleder, Natalia Higuita-Castro*
[1]*Department of Physiology and Cell Biology, College of Medicine, The Ohio State University, Columbus, Ohio, United States*

SP48 Dual AAV and split intein strategy for functional dysferlin restoration in LGMDR2

Nikki M. McCormack, Stephanie Lee-Diaz, Theodore R Reyes, Marshall W. Hogarth, James S. Novak, Christine L. Halbert, Jeffrey S. Chamberlain, David L. Mack, Martin K. Childers, Jyoti K. Jaiswal, ***Abdeslem Hichem Tasfaout***[1]
[1]*University of Washington, Seattle, United States*

Short Oral Presentations 12: SP49-SP53, Novel approaches in NMD

SP49 A proteoglycan serglycin promotes skeletal muscle regeneration through EZH2 phosphorylation in myoblasts

Katsuhiko Kunitake[1,2], *Yuki Ashida, Norio Motohashi, Yoshitsugu Aoki*
[1]*Department of Neurology, Fujita Health University School of Medicine, Toyoake, Japan,* [2]*Department of Molecular Therapy, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Kodaira, Japan*

SP50 Desmin undergoes active nuclear translocation in human skeletal myoblasts and functions as a dynamic nucleo-cytoskeletal scaffold

Nilüfer Düz[1], *Ecem Kural-Mangıt, Ahmet Avcı, Onur Aktan, Şevval Şahin, Süreyya Özcan, Turgut Baştuğ, Pervin Dinçer*
[1]*Hacettepe University, Ankara, Turkey*

SP51 Delivery of therapeutic RNAs to skeletal muscle using lipid nanoparticles

Cecilia Jiménez-Mallebrera[1,2], *Baris Ongoren, Aristides López-Márquez, LLuis Enjuanes, Mariana Köber, Carmen Badosa*
[1]*Institut de Recerca Sant Joan De Déu, Barcelona, Spain,* [2]*CIBERER, ISCIII, Madrid, Spain, 3Instituto de Ciencias Materiales de Barcelona-CSIC, Barcelona, Spain*

SP52 Lentiviral optogenetic engineering of human 3D neuromuscular junctions for disease modeling applications

Nelsa Estrella[1], *Jacob Fleming, Thomas Leahy, Hamed Ghazizadeh, Mohammad Khoonkari, Christal Worthen, Greg Luerman, Nicholas Geisse*
[1]*Curi Bio, Seattle, United States*

SP53 Pen bioprinting of iPSC-derived muscle progenitors promotes functional skeletal muscle regeneration

Yori Endo[1], *Indranil Sinha, Olivier Pourquié, Ali Tamayol*
[1]*Mass General Brigham, Harvard Medical School, Boston, United States*

Saturday 3 October 2026

Clinical trial updates: 17O – 22O

17O RGX-202, a phase I/II/III investigational gene therapy for the treatment of Duchenne muscular dystrophy

*Steve Pakola, Aravindhan Veerapandiyan, Crystal Proud, Nancy Kuntz, Amy Harper, Susan Iannaccone, **Carolina Tesi-Rocha**[1], Erika Finanger, Cuixia Tian, Han Phan, Eleonora D'ambrosio, Dimah Saade, Jahannaz Dastgir, Regenxbio RGX-202 Study Team, Koji Takahashi, Olivier Danos*

[1]Stanford School of Medicine, Stanford, United States

18O HOPE-3, a phase 3 study of deramiocel, an allogeneic cell therapy, in advanced Duchenne muscular dystrophy: evidence to support both musculoskeletal and cardiac efficacy

Craig McDonald[1], *Chet Villa, Jon Soslow, Erik Henricson, Cuixia Tian, Russell Butterfield, Kati Maharry, Nathan Hogan, Michael Binks, Kevin Berth, Mark Awadalla, Eugenio Mercury, Eduardo Marban, Linda Marban*

[1]University of California Davis, Sacramento, United States

19O Long-term safety and efficacy of resamirigene bilparvovec for the treatment of x-linked myotubular myopathy (ASPIRO)

Nancy Kuntz[1], *Rahul Gentyala, Julie Coats, Atsuki Hashimoto, Astrid Blaschek, Wolfgang Muller-Felber, Carston G. Bonnemann, Barbara K. Smith, Andrea Sefarian, Lucy James, Perry B. Shieh, James J. Dowling*

[1]Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, United States

20O Epigenetic silencing in FSHD: preliminary clinical safety and efficacy of the novel gene therapy EPI-321

Richard Roxburgh[1], *Miriam Rodrigues, Renata Shih, Han Phan, Russell Buttterfield, Doris Leung, Lawrence Hayward, Catherine Douthwright, Katrina Morris, Zaki Jawaid, Vishi Agarwal, Andrew Norton, Nalinda Wasala, Abhinav Adhikari, Alexandra Collin de l'Hortet, Daniel Hart, Dipali Patel, Kavita Jadhav-Whalen, Weston Miller, Perry Shieh*

[1]Neurogenetics Research Clinic, Centre for Brain Research, University of Auckland, Auckland, New Zealand

21O Interim analysis from the ongoing Phase 3 FORTIFY study of BBP-418 for individuals living with LGMD2I/R9 suggests benefit across functional and cardiac endpoints

*Damon Bass, Katherine D. Mathews, Tahseen Mozaffar, **Volker Straub**[1], Robert D. Henderson, Douglas Sproule, Ada Lee, Amy Rainey, David Paul, Tricia Blankenbiller, Uma Sinha, John Vissing, for the FORTIFY study group*

[1]Newcastle University, United Kingdom

22O Oral salbutamol as adjuvant therapy in generalized myasthenia gravis: a randomized controlled trial

Josefine Jul Jarbæk Nielsen[1], *Jan Lykke Scheel Thomsen, Izabella Obál, Jakob Vormstrup Holbech, Henning Andersen*

[1]Neurology, Aarhus University Hospital, Aarhus, Denmark

Virtual Posters

1.001VP Phase 1 study of a PROTAC polyQ-AR degrader (ARV-027) in healthy participants and participants with spinal and bulbar muscular atrophy

James Gregory[1], *Jiachang Gong, Miri Alon, Johnathan Obourn, Kyle Fraser, Madeleine Ratte, Ilaria Conti, Jochem Stormmesand, Andriy Gorbenko, Philip Kremer*

[1]*Arvinas Operations, Inc., New Haven, United States*

1.002VP TTN splicing variant effect detection on variants at transcriptome level

Victoria Lillback[1], *Swethaa Natraj Gayathri, Ali Oghabian, Maria Francesca Di Feo, Mridul Johari, Peter Hackman, Bjarne Udd, Marco Savarese*

[1]*Folkhälsan Research Center, Helsinki, Finland*

1.003VP Presymptomatic risdiplam in preterm monozygotic twins with co-occurring spinal muscular atrophy and tuberous sclerosis complex

Xiaofang Zhang[1], *Yanjuan Wang, Sixiu Li, Ping Liu, Wenguang Hu*

[1]*Chengdu Women and Children's Center Hospital, Chengdu, Sichuan Province, China, Sichuan Province, China*

1.004VP Assessment of general movements in neuromuscular disorders: a scoping review

Zeynep Arıkan[1], *Fatma Uğur*

[1]*Karamanoglu Mehmetbey University, Karaman, Turkey*

1.005VP Utility of motor unit number index for assessment of disease progression in Hirayama disease

Sruthi Sivaraman Nair[1], *Sri Sai Srujana Puppala, Jemini George, Karthika Ajit V*

[1]*Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, India*

2.001VP. Early cardiac involvement in children with Duchenne muscular dystrophy

Kavitha Prasad[1,2], *Monika Sharma, Vineeth Jaison, Gurbhej Singh, Anmol Bhatia, Megha Khanna, Karun Khanna*

[1]*Christian Medical College , Ludhiana, India,* [2]*Department of Pediatrics Christian Medical College , Ludhiana , India*

2.002VP Correlation between cardiovascular, respiratory and surface electromyography findings with functional tests in non-ambulatory patients Duchenne muscular dystrophy

Rosa Elena Escobar Cedillo[1], *Jose Francisco Ramos Becerril, Cecilia Alejandra Rodríguez Chávez, Cinthia Lourdes Toledo Peral, Gabriel Vega Martinez, Sonia Patricia Romano Riquer, Nataly Margarita Nuñez Hernández*

[1]*Instituto Nacional De Rehabilitación, Ciudad De México, Mexico*

2.003VP Unravelling DMD duplications with targeted long-read sequencing

Alba Segarra-Casas[1], *Anna Delgado-Tejedor, Maik Zehnsdorf, Anna Menoyo, Maria José Rodriguez, Olaya Villa, Clara Serra, Ivon Cuscó, Julia Ponomarenko, Pia Gallano, Lidia Gonzalez-Quereda, Berta Fusté*

[1]*Institut de Recerca Sant Pau (IR SANT PAU), Barcelona, Spain*

2.004VP Neurodivergence in Duchenne muscular dystrophy capturing the "big 10": from an Indian cohort

Nelia Mathew[1], *Himashweta Balwani, S Vedha Reddy, Ann Mathew*

[1]*St Johns Medical College Hospital, Bengaluru, India*

3.001VP Late-onset Pompe disease adverse event management: a consensus for patient safety

Olimpia Musumeci[1], *Lucia Ruggiero, Guido Alessandro Primiano, Michele Croce, Giuseppina Miele, Simona D'Amore, Anna De Nes, Barbara Risi, Giulia Ricci, Cristina Sancricca, Liliana Vercelli, Federico Spadaro*

[1]*University of Messina, Messina, Italy*

3.002VP PX578: a first-in-class POLG activator to increase mtDNA for treatment of mitochondrial DNA depletion syndromes

Juli Jones[1], *Xie Xie, Courtney Heard, Sofie Ekström, Barbara Küppers-Munther, S. Jordan Kerns, Laleh Arabanian, Carol Geukens, Anja Gruszczyk, Benjamin Munro, Camilla Koolmeister, Nils-Goran Larsson, Claes Gustafsson, Maria Falkenberg, Simon Giroux, Andrew Griffin, Thomas Keating, Gabriel Martinez Botella*

[1]*Pretzel Therapeutics, Waltham, United States*

4.001VP Clubfoot in KLHL41-related congenital nemaline myopathy: a rare case report

Ana Cotta[1], *Frederico Godinho, Elmano Carvalho, Antônio-Lopes Cunha-Júnior, Alexandre Cauhi, Eni da-Silveira, Cynthia Costa-e-Silva, Jaquelin Valicek, Julianne Lima, Brunno Moreira*

[1]*The Sarah Network of Rehabilitation Hospitals, Belo Horizonte, Brazil*

4.002VP Herpesvirus viral load and interferon dysregulation in acetylcholine receptor-positive myasthenia gravis

Ieva Glāzere[1,2], *Arta Grosmane- Bataraga, Vladimirs Krutovs, Marija Roddate, Gundega Ķauķe, Linda Gailīte, Anda Vilmane, Lība Sokolovska, Maksims Zolovs, Kai Kisand, Nataļja Kurjāne, Viktorija Ķēniņa*
[1]*Department of Neurology, Pauls Stradiņš Clinical University Hospital, Riga, Latvia,* [2]*Department of Biology and Microbiology, Riga Stradiņš University, Riga, Latvia*

4.003VP Patient reality vs scientific priorities in DM1: a multi-source comparative study using the French DM-scope registry

Melinda Gyenge[1], *Manon Brechenmacher, Frédéric Fer, Dalil Hamroun, Gauillaume Bassez, French Filnemus Myotonic Dystrophy Study Group*
[1]*Institute of Myology, Pitié Salpêtrière Hospital, Paris, France*

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